

Mosaic ectodermal epigenetic dysregulation in autism spectrum disorder.Berko *et al.***Table of contents:**

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Patient Cohort

All patient recruitment and sample collection was done with the appropriate human subjects protocol approval from the Institutional Review Board at the Albert Einstein College of Medicine.

We enrolled two groups of subjects: descriptive traits of the two groups are summarized in **Supplemental Table S1**.

We recruited subjects with an ASD by two primary methods: enrolling patients seen in our clinics, and recruiting patients nationally from advertisements and a posting on the Autism Speaks website (<http://www.autismspeaks.org>). Patients enrolled from our clinics and research labs (43 subjects) were diagnosed with an ASD by combinations of metrics including the ADOS, ADI-R, CARS, DSM-IV, and clinical assessment, by clinicians and researchers trained in ASD diagnostic measures.

Other subjects enrolled in our study by internet and external recruitment (7 subjects) provided reports of an ASD diagnosis by ADOS and/or ADI-R from an accredited institution with one exception, a patient diagnosed with an ASD by the New York City Early Intervention Program.

The control cohort consisted of typically developing individuals without any prior evidence of an autism spectrum disorder.

| | ASD | TD |
|-------------------------|--------------|--------------|
| Ages | | |
| Maternal age (mean, SD) | 37.58, 2.93 | 38.1, 2.97 |
| (range) | (35-48) | (35-48) |
| Paternal age | 40.19, 6.27 | 40.72, 5.87 |
| | (29-51) | (30-52) |
| Subject Age | 6.84, 3.58 | 11.2, 7.31 |
| | (2-17) | (1-28) |
| Gender | | |
| Male (n) | 39 | 22 |
| Female | 11 | 28 |
| Genetic Ancestry | | |
| % CEU (mean, SD) | 63.38, 29.37 | 85.19, 16.38 |
| % YRI | 22.43, 30.74 | 6.61, 17.02 |
| Total | 50 | 50 |

Supplemental Table S1: Characteristics of study subjects.

Age metrics reflect the mean age in years per group and the standard deviation, with the range included in parentheses. Maternal and paternal age refer to the age of the parents at the time of the subject's birth. All maternal ages are included, while information for paternal age was only available for ~50% of subjects. Subject age refers to age when the sample was collected. Percent genetic ancestry based on subjects with quality filtered genotype data; 47 ASD and 46 TD genotypes were included.

ASD: Autism Spectrum Disorder.

TD: Typically Developing.

CEU: Utah residents with Northern/Western European ancestry from the CEPH collection (European)

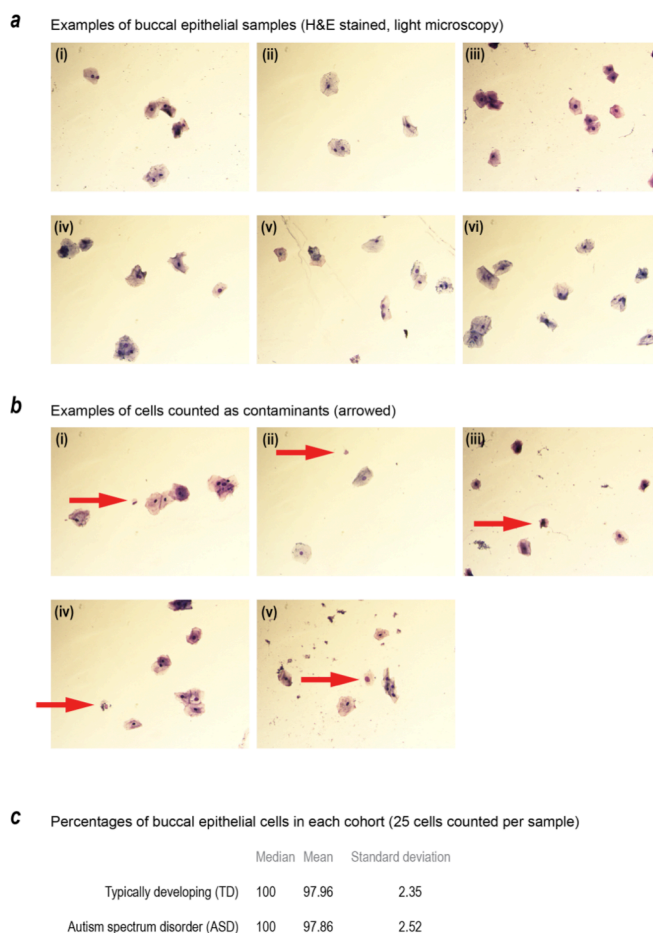
YRI: Yoruba in Ibadan, Nigeria (African)

Sample Collection

We optimized a method to collect DNA from buccal epithelium, using brushes to exfoliate the cheek and cheek gutters (area between the upper/lower gumline and cheeks). Brushes were then placed in CytoLyt (Hologic, MA), a methanol-based preservative. Upon arrival in the laboratory, brushes were removed from the solution, and cell pellets were retrieved from the sample, snap frozen in liquid nitrogen, and moved to -80°C for long term storage.

Cell subtype composition was determined by preparing slides from each of the samples collected, performing haematoxylin and eosin (H&E) staining, and visualizing using light microscopy, counting the first 25 individual (not clumped) cells seen. The typical appearance of these cells plus examples of cell-like structures counted as contaminants in **Supplemental Figure S1a-b**. Comparably high purity of the squamous epithelial cells was found for both groups.

We performed DNA extraction from these cells and confirmed the presence of high quality and high molecular weight DNA with UV spectrometry and gel electrophoresis.



Supplemental Figure S1: H&E staining of buccal epithelial samples.

The buccal brushings yielded the expected squamous epithelial cells (a) with very small proportions of possible contaminant cells types (b) of different sizes (b.i-b.iv) or nuclear characteristics (b.v). Counts of 25 individual cells per sample showed high purity in both ASD and TD groups (c).

Genotyping and Methylation microarrays

For the epigenome-wide association study (EWAS), we used a microarray-based approach, as the presence of oral bacterial DNA in the extracted samples precluded cost-effective sequencing.

Samples were randomized across arrays, and plates were submitted in batches of 96 to minimize potential for batch effect. Sufficient DNA was extracted to meet manufacturer's requirements of 200 ng and 500 ng for the genotyping and methylation arrays, respectively. Microarray hybridization and subsequent scanning was performed by a core facility according to manufacturer's protocols. The summary of samples run on each microarray is provided in **Supplemental Table S2**.

The Illumina HumanOmni2.5-8 BeadChip genotyping platform was used for genotyping, and the Illumina Infinium HumanMethylation450 Beadchip platform for DNA methylation studies.

| | ASD | TD | Parents |
|-------------------------------|-----|------------------------------|---------|
| <i>Genotyping microarrays</i> | | | |
| Batch 1 | 24 | 24 (including 2 siblings) | 47 |
| Batch 2 | 26 | 26 (including 5 siblings) | 45 |
| <i>Methylation microarray</i> | | | |
| Batch 1 | 47 | 48 | 0 |

Supplemental Table S2: Experimental batches of microarrays

Genotyping Data Preprocessing

Raw data from the genotyping microarrays were loaded into *BeadStudio*, Illumina's proprietary chip analysis software. Genotype calls were then exported in *plink* format, specifying conversion to the forward strand, which refers to annotation based on dbSNP.

We used *plink* to filter the data, with the following parameters for missingness and allele frequency:

```
--mind 0.05 --geno 0.1 --maf 0.01 --hwe 0.001
```

Filtering retained 184 out of 191 individual arrays, and 1,777,521 out of 2,379,855 SNPs, with genotyping rate of 0.992 in these remaining samples.

We then removed all ambiguously mapping single nucleotide polymorphisms (SNPs), designated by Illumina as "chr0".

Since we wanted to perform local ancestry deconvolution based on the 1000 Genomes data, which by convention is annotated on the "+" strand of hg19, we had to ensure our Illumina data likewise all mapped to the "+" strand. We therefore identified which of the dbSNP polymorphisms represented on the array actually mapped to the "-" strand, and used *plink* to flip those bases exclusively. We subsequently further removed the handful of SNPs on each chromosome that still possessed two variant alleles. We confirmed correct strandedness by merging our genotyping data with 1000 Genomes data and performing principal components analysis using *vcftools* (Danecek et al. 2011).

Local Ancestry Deconvolution

Since principal components analysis of our dataset revealed many individuals of mixed 3-way genotype admixture, we chose to perform local ancestry deconvolution with simulated mixed ancestry using *HapMix*, an approach previously shown to be accurate (Price et al. 2009). *HapMix* is an algorithm which uses a hidden Markov model to estimate the probability of ancestry at each data point along a chromosome, based on the input of 2 parental ancestral populations and rates of recombination. To model 3-way admixture, we ran two iterations of *HapMix* with different mixed parental populations, and calculated the joint probability of homozygous or heterozygous genotypes at each point (Price et al. 2009).

As in Price *et al.*, we constructed 2 sets of reference populations using genotype data from the 1000 Genomes project from Caucasians (CEU), Africans (YRI) and East Asians (CJ: CHB + JPT). After randomly selecting 80 unrelated individuals from each group, we constructed two mixed sets: a CEU/(CHB+JPT) set with 40 individuals from each group (80 total), and a CEU/YRI set with 40 individuals from each group (also 80 total). We then ran *HapMix* twice for each chromosome; first with the 2 parental populations as the CEU/(CHB+JPT) mixed set and homogeneous YRI, and then with the 2 parental populations as the CEU/YRI mixed set and homogeneous (CHB+JPT).

For each HapMix run, at every locus, *HapMix* estimates the probability of a homozygous call of the genotype of parental population 1, a heterozygous call of one allele from each parental population, and a homozygous call of the genotype of parental population 2. To calculate the final joint probability from both runs, we employed the following calculations (Price et al. 2009), explained in **Supplemental Table S3**:

RUN 1: CEU/(CHB+JPT) versus YRI as the two potential parental populations

RUN 2: CEU/YRI versus (CHB+JPT) as the two potential parental populations

The probabilities of the six potential states at each locus were normalized to 1, and final ancestry calls were made as the maximum probability at the locus only if it exceeded 0.5.

| Call | Probability Calculation (run number) |
|----------------------|---|
| Homozygous YRI | Probability YRI (from run 1) X Probability CEU/YRI (from run 2) |
| Homozygous CJ | Probability CEU/(CHB+JPT)(1) X Probability (CHB+JPT)(2) |
| Homozygous CEU | Probability CEU/(CHB+JPT)(1) X Probability CEU/YRI(2) |
| Heterozygous YRI/CJ | Probability heterozygous CEU/(CHB+JPT) & YRI(1) X Probability heterozygous CEU/YRI & (CHB+JPT)(2) |
| Heterozygous YRI/CEU | Probability heterozygous CEU/(CHB+JPT) & YRI(1) X Probability CEU/YRI(2) |
| Heterozygous CJ/CEU | Probability CEU/(CHB+JPT)(1) X Probability heterozygous CEU/YRI & CJ(2) |

Supplemental Table S3: *HAPMIX* local ancestry probability calculation

Abbreviations:

CHB: Han Chinese in Beijing, China.

JPT: Japanese in Tokyo, Japan

CJ: CHB/JPT (East Asian)

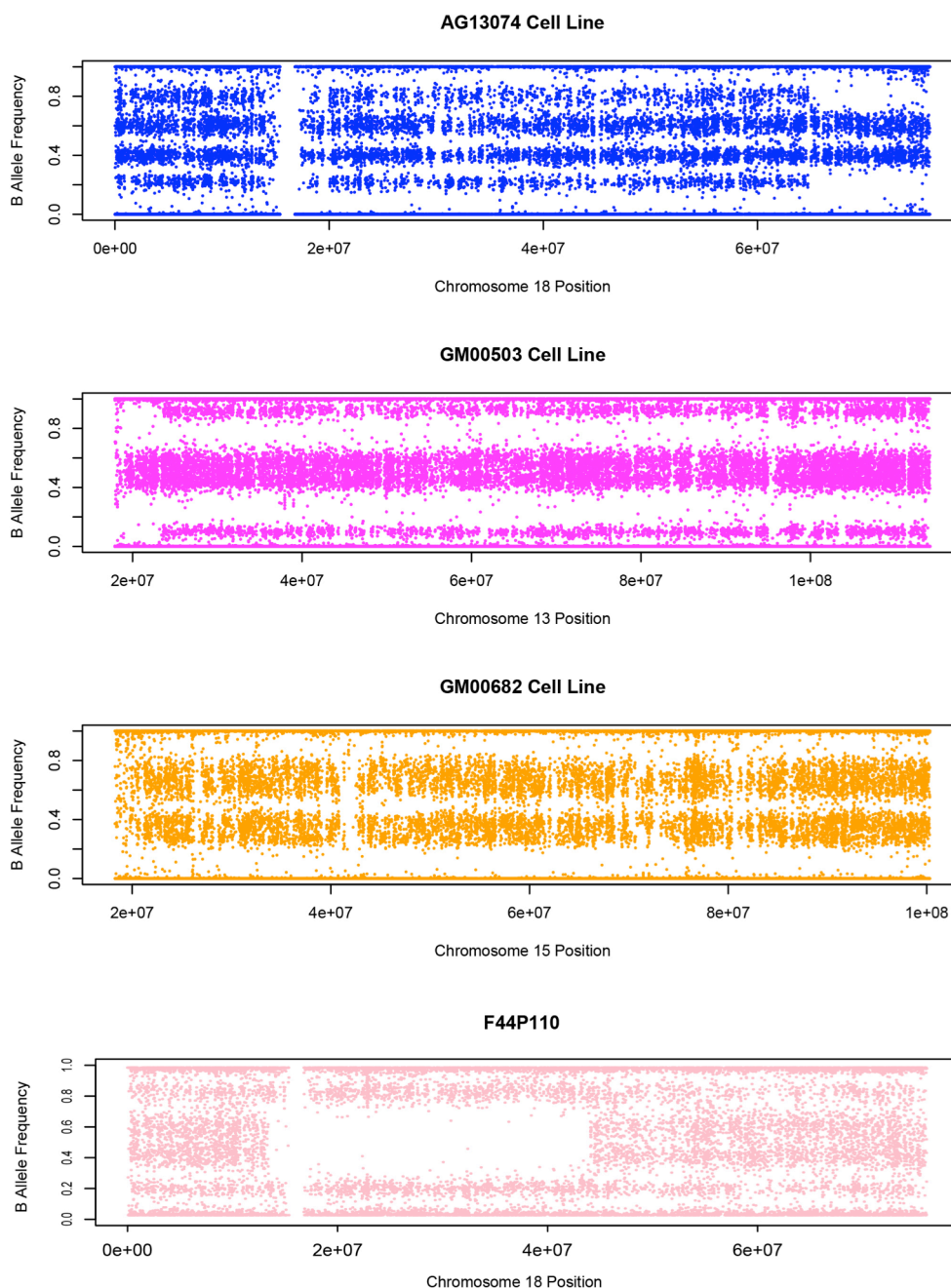
YRI: Yoruba in Ibadan, Nigeria (African)

CEU: Utah residents with Northern and Western European ancestry from the CEPH collection (European)

Mosaicism Detection

We used the *Mosaic Alteration Detection (MAD)* algorithm, implemented in the package *GADA*, to identify potential chromosomal mosaic events (Gonzalez et al. 2011). To confirm our ability to use *MAD* to discover detectable whole chromosome mosaicism, we prepared a test dataset of 4 samples. The samples consisted of DNA from 3 cell lines, known to harbor mosaic trisomy, from Coriell Cell Repository, and one DNA sample extracted from buccal epithelium of a patient with a clinical diagnosis of mosaic trisomy 18. Samples were hybridized on the Illumina Infinium Omni1-Quad (prior to Illumina's release of the 2.5-8 BeadChip) and genotype calls were made with Illumina Genome Studio. We plotted the B-Allele Frequencies (BAF) of each known mosaic chromosome (**Supplemental Figure S2**) confirming the presence of visually detectable mosaic trisomy. From the BAF plots we were further able to characterize the mosaicism as resulting from either a meiosis I or meiosis II non-disjunction, based on loss of the 5th and 6th genotype bands at either the telomere or centromere, respectively. We were also able to identify segments of uniparental disomy (UPD), based on loss of heterozygous bands (Conlin et al. 2010). We then analyzed the genotypes with *MAD*, employing the default suggested parameters ($\alpha=0.8$, $T=9$, and $\text{MinSegLen}=75$). Although it also called mosaic events on other chromosomes, *MAD* correctly identified the whole chromosome mosaic trisomy in all 4 cases, either with an over-abundance of mosaic calls or by calling exceptionally long mosaic regions on the affected chromosome.

To prepare our genotyping dataset for *MAD* analysis, we first re-clustered the data in GenomeStudio to improve the quality of some poorly performing SNPs. After exporting the data and applying the same *plink* filters shown previously, we retained 186 out of 191 individual arrays and 1,820,947 SNPs. We ran *MAD* with the same default parameters as our test dataset, obtaining 376 called mosaic segments across the 186 individuals. No whole chromosome mosaicism events were detected in either the ASD or the TD subjects.



Supplemental Figure S2: B allele frequency plots of known mosaic cases

The *MAD* output correctly highlights an abnormality along chromosomes; analysis of BAF patterns allows determination of the source of error. The pattern in AG13074 results from meiosis I non-disjunction, and F44 P110 demonstrates a meiosis II non-disjunction with mosaic UPD. The lack of array probes on the p arm of chromosome 13 precludes definitive assessment of the meiotic source of the GM00503 error (I or II); the pattern of GM00682 could result from a variety of situations.

Methylation Data Preprocessing

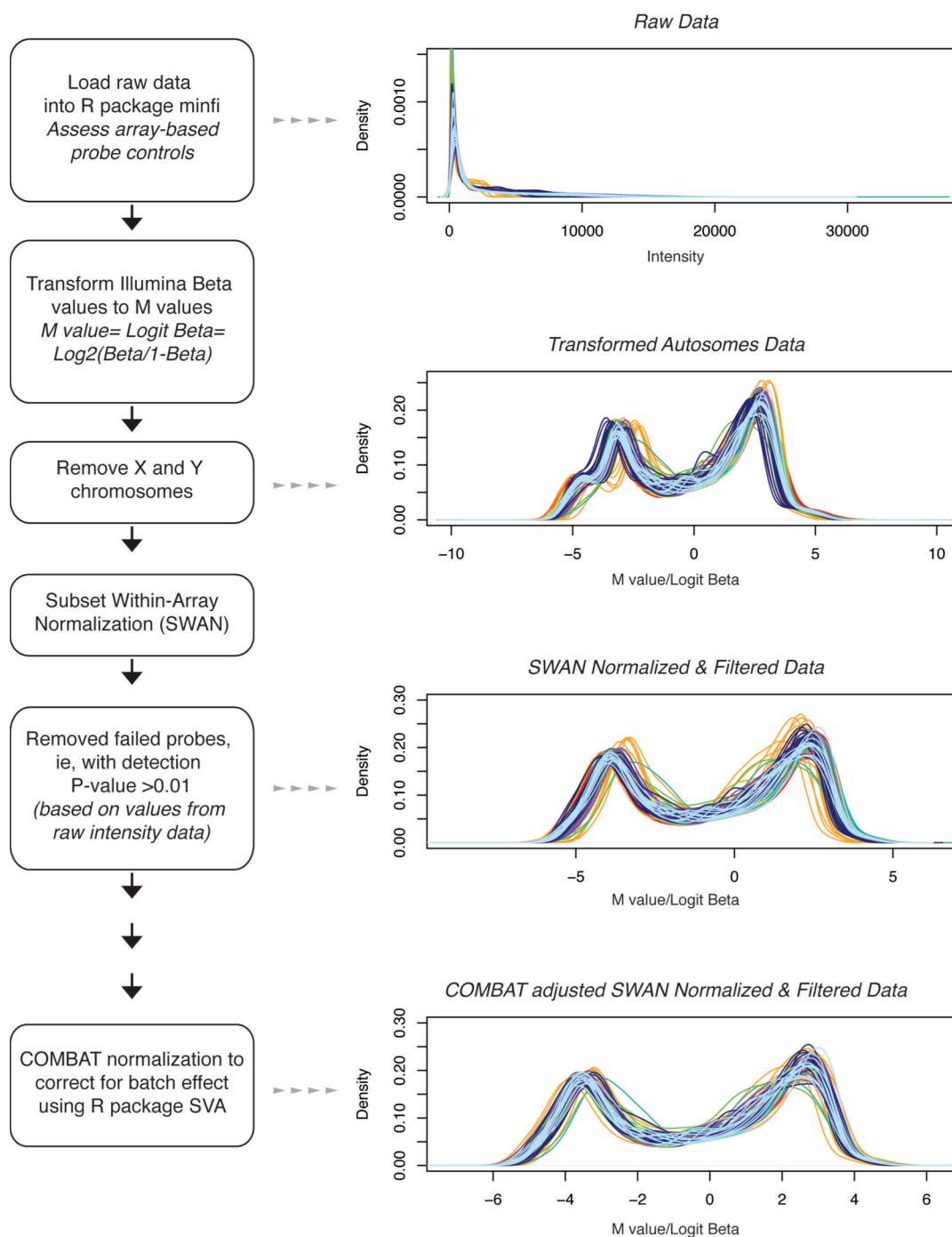
Supplemental Figure S3 provides an overview of the preprocessing pipeline.

Raw *idat* data were loaded in the R (RCoreTeam 2012) package *minfi* (Hansen and Aryee) and assessed for basic quality control metrics, including determination of poorly performing probes with insignificant detection p-values above background control probes. Since our cohort includes both males and females, we removed the X and Y chromosomes from the raw methylation values, and performed *SWAN* normalization to correct for intra-array differences between Illumina Type I and Type II probes (Dedeurwaerder et al. 2011; Maksimovic et al. 2012). We excluded any probes with detection p-value greater than 0.01 (12,590 probes), and then corrected for batch (microarray chip) effect using the *ComBat* function in the R package *SVA* (surrogate variable analysis) (Leek et al.).

Differentially-methylated region (DMR) Identification

To understand the relative effects of known technical, biological, and microarray-based covariates acting on methylation data variability, we performed principal components analysis (PCA) on the M values (logit-transformed Illumina-defined beta values) obtained from the previous preprocessing. We accounted for the possible known confounders, including technical (date of DNA extraction, microarray chip, position on chip), microarray-based (all categories of control probes designed by Illumina) and biological (ASD status, age, gender, and ancestry percentage). Ancestry percent was calculated as the proportion for each population of all allele genotyping positions called by *HapMix*. We fit a linear model for each of the top 10 principal components as a function of each covariate, and summarized the data with a heatmap of the negative \log_{10} p-values for each regression. We identified the significant confounding covariates and corrected for them in all subsequent analysis.

Bump-hunting provides significant advantages over typical individual probe statistical test: it smoothes data over a region, obliterating the need for arbitrary genomic cutoffs, it applies a rigorous analysis and correction for confounding factors, and it incorporates false discovery rate (FDR) considerations as part of the algorithm. Bump-hunting utilizes known covariates input into a model matrix as components in regression analyses. Although we did not see a strong effect of gender on PCA variability, we included gender in our model matrix since methylation patterns are known to vary by gender (Sarter, Long et al. 2005). Based on our PCA data, we input age, percent CEU (European), and percent YRI (African) ancestry as known biological covariates. Bump-hunting was used in the *dmrFind* function in the R package *charm* (Aryee et al. 2011).

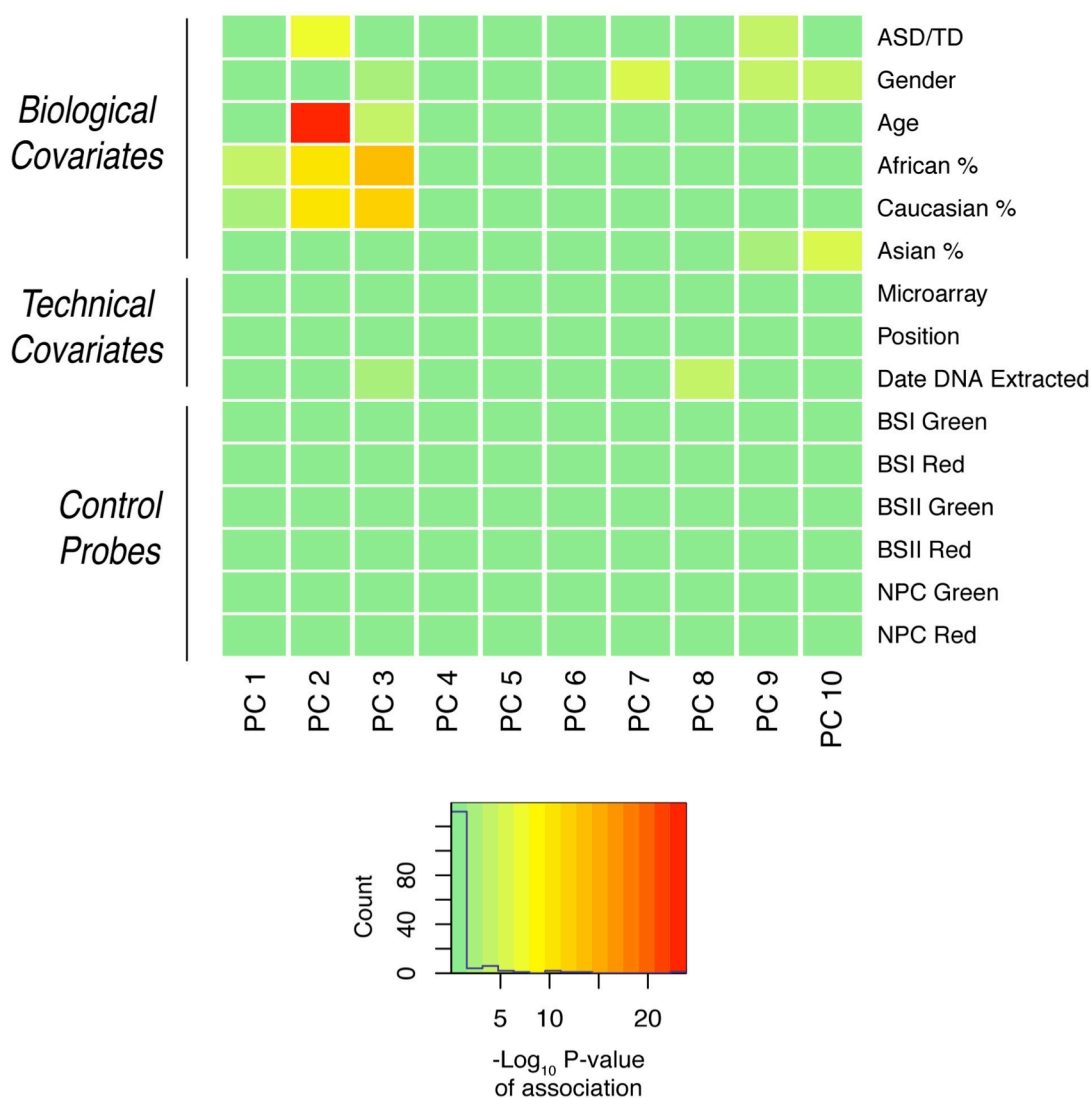


Supplemental Figure S3: Preprocessing of Illumina 450K data

The colors in the density plots correspond to different chips (microarrays), each run to contain 12 samples. The normalization procedures correct for both intra-array and inter-array differences.

We excluded two TD samples from our analysis, since the genotyping arrays performed on their DNA did not pass quality control thresholds.

The *dmrFind* algorithm corrects for unidentified confounders, and returns probe methylation values that preserve the effects of the known covariates input into the model matrix. PCA and linear regression on this data confirmed that SVA properly identified and corrected for all unwanted technical sources of variation, **Supplemental Figure S4**.



Supplemental Figure S4: Associations between principal components and known covariates

Heatmap of $-\log_{10}$ P-values for the association of each principal component with each known covariate demonstrates that variation due to technical artifact has been removed, while variation due to known biological covariates has been preserved for subsequent analysis.

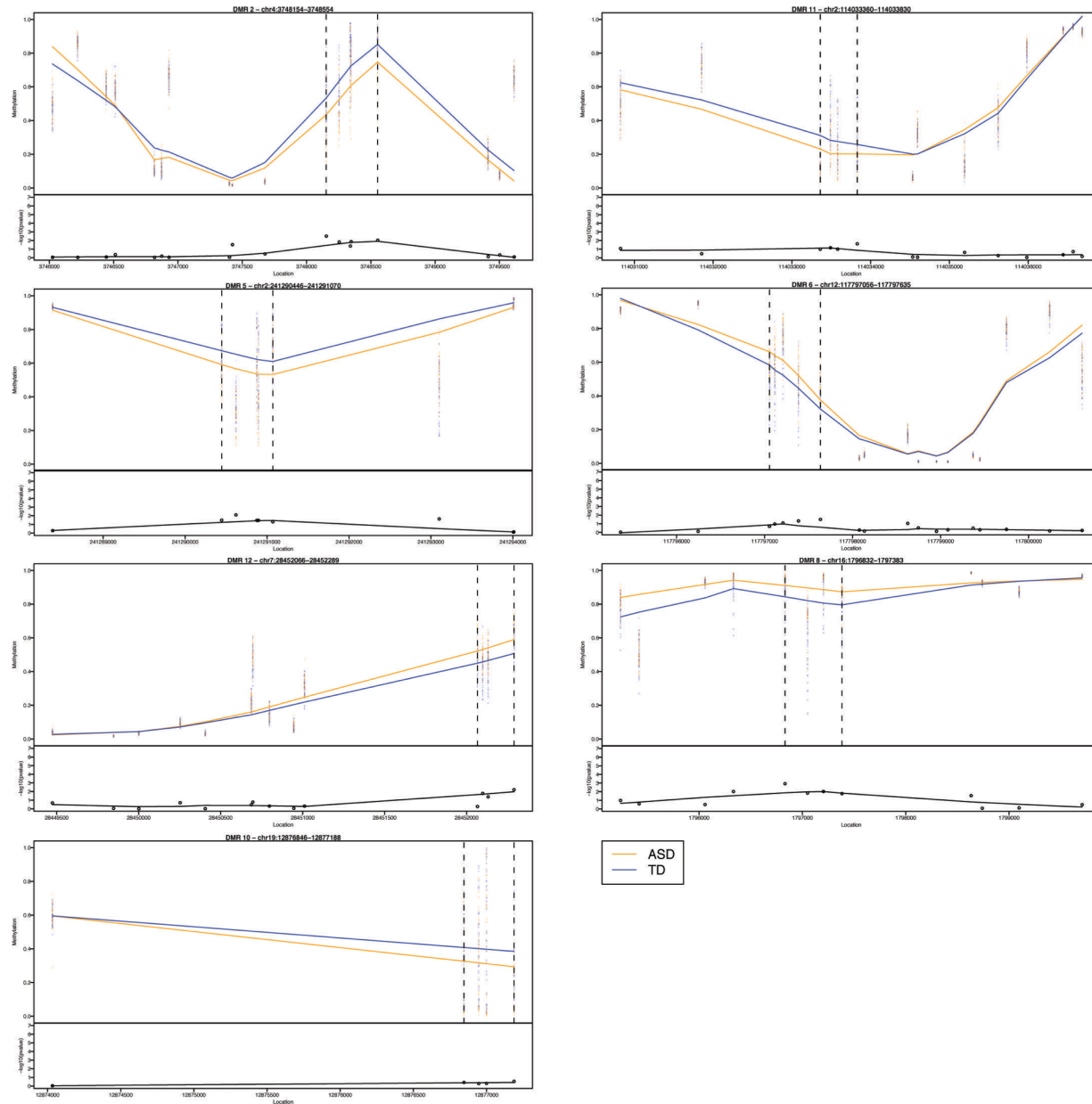
The bump-hunting ASD-associated DMR output is provided in **Supplemental Table S4**. Stable DMRs are visualized in **Supplemental Figure S5**.

The differentially methylated regions were associated with the nearest overlapping gene, shown in **Table 1**. We investigated autism gene databases (Xu et al. 2012) and published literature for previous evidence linking the gene to ASD. We used UCSC genome browser (Kent et al. 2002) to determine the cytoband, and the SFARI gene CNV database (Basu et al. 2009) for a summary of CNV research implicating the cytoband in ASD. Gene functions were gleaned from UCSC genome browser and the GeneCards database (Rebhan et al. 1997).

| chromosome | start | end | value | area | pns | indexStart | indexEnd | nprobes | avg | max | area.raw | gene name | illumina annotation |
|------------|-------------|-------------|--------|-------|---------|------------|----------|---------|--------|--------|----------|------------------|--|
| 1 | 248,100,183 | 248,100,614 | 0.427 | 4.273 | 23,221 | 45,365 | 45,374 | 10 | 0.080 | 0.150 | 0.803 | <i>OR2L13</i> | 1st exon, 5'UTR, TSS200 and TSS 1500, CpG island and north shore |
| 2 | 241,290,446 | 241,291,070 | 0.381 | 1.904 | 137,222 | 274,789 | 274,793 | 5 | 0.086 | 0.104 | 0.428 | <i>GPC1</i> | north shelf |
| 2 | 114,033,360 | 114,033,830 | 0.331 | 1.323 | 128,333 | 258,706 | 258,709 | 4 | 0.075 | 0.105 | 0.301 | <i>PAX8</i> | gene body, CpG island and north shore |
| 4 | 3,748,154 | 3,748,554 | 0.465 | 2.324 | 163,348 | 326,311 | 326,315 | 5 | 0.119 | 0.148 | 0.595 | <i>ADRA2C</i> | CpG island and north shore |
| 5 | 139,227,979 | 139,228,242 | -0.158 | 0.790 | 180,529 | 358,423 | 358,427 | 5 | -0.076 | -0.127 | 0.378 | <i>NRG2</i> | gene body, CpG island |
| 5 | 16,508,920 | 16,509,123 | 0.373 | 1.491 | 174,420 | 347,241 | 347,244 | 4 | 0.078 | 0.095 | 0.312 | <i>FAM134B</i> | TSS200, 5'UTR, 1st exon, body, enhancer |
| 6 | 73,329,988 | 73,330,358 | -0.209 | 1.253 | 192,434 | 389,727 | 389,732 | 6 | -0.077 | -0.088 | 0.461 | <i>KCNQ5</i> | TSS1500, CpG island north shore |
| 7 | 28,452,066 | 28,452,289 | -0.202 | 0.806 | 202,381 | 410,259 | 410,262 | 4 | -0.076 | -0.083 | 0.302 | <i>CREB5</i> | TSS200, 5'UTR, 1st exon, CpG island south shelf |
| 10 | 135,341,870 | 135,342,620 | -0.491 | 2.947 | 35,126 | 69,284 | 69,289 | 6 | -0.078 | -0.148 | 0.469 | <i>CYP2E1</i> | gene body, CG island and south shore |
| 10 | 135,342,936 | 135,343,280 | -0.462 | 1.850 | 35,127 | 69,290 | 69,293 | 4 | -0.078 | -0.098 | 0.310 | <i>CYP2E1</i> | gene body, south shore |
| 12 | 117,797,056 | 117,797,635 | -0.213 | 1.067 | 57,778 | 115,485 | 115,489 | 5 | -0.086 | -0.109 | 0.432 | <i>NOS1</i> | 5'UTR, north shore |
| 16 | 53,407,678 | 53,407,808 | 0.174 | 0.696 | 87,732 | 174,199 | 174,202 | 4 | 0.089 | 0.115 | 0.357 | <i>LOC643802</i> | CpG island, south shore |
| 16 | 2,879,944 | 2,880,326 | -0.265 | 1.060 | 83,603 | 166,459 | 166,462 | 4 | -0.101 | -0.125 | 0.402 | <i>ZG16B</i> | TSS200, TSS1500, 1st exon |
| 16 | 1,796,832 | 1,797,383 | -0.515 | 2.060 | 83,090 | 165,290 | 165,293 | 4 | -0.090 | -0.139 | 0.361 | <i>MAPK8IP3</i> | gene body, CpG island and south shore |
| 19 | 12,876,846 | 12,877,188 | 0.401 | 1.603 | 111,948 | 225,403 | 225,406 | 4 | 0.079 | 0.086 | 0.315 | <i>HOOK2</i> | gene body, CpG island, north shore and south shore |

Supplemental Table S4: DMRs Associated with ASD from Bump-Hunting.

Unstable DMRs are shaded in gray.



Supplemental Figure S5: Stable DMRs defined by the *bump-hunting* algorithm

DNA methylation values are displayed along with $-\log_{10}$ p-values for each probe.

Left column, from top to bottom, gene names: *ADRA2C*, *GPC1*, *CREB5*, *HOOK2*.

Right column, top to bottom: *PAX8*, *NOS1*, *MAPK8IP*.

Copy number variant (CNV) Calling

The *CNVision* algorithm (Sanders et al. 2011) integrates three CNV-calling platforms: *GNOSIS*, *PennCNV*, and *QuantisNP* (Colella et al. 2007), increasing accuracy of calls with this complementary assessment of each CNV. Since we wanted to exclude any loci with potential CNVs from methylation analysis, we retained all called CNVs, even those called with low potential probability.

When DMRs were found to overlap CNVs, we re-ran bump-hunting with these loci excluded to confirm that the local DMR was not solely due to the presence of CNVs in the region.

Massively-parallel bisulfite sequencing to validate differential DNA methylation

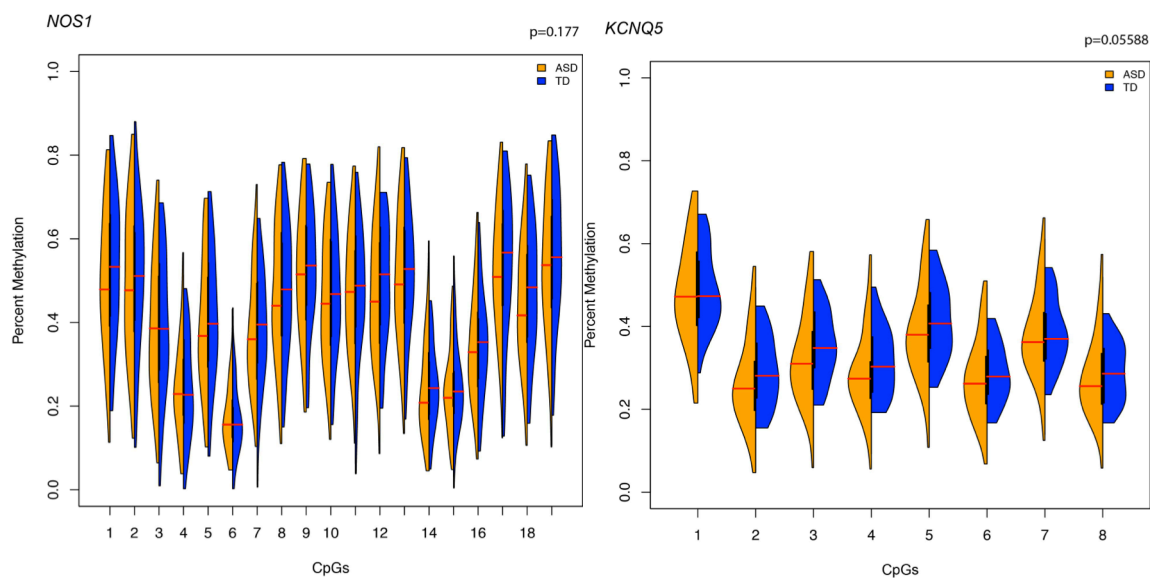
We bisulphite converted 500 ng of DNA using the Zymo EZ-96 Methylation-Lightning Kit. After separate PCR amplification of 4 target regions, we pooled the amplicons in averaged equal ratios and generated Illumina libraries using Tecan automation. Two sets of 48 libraries each were multiplexed on the MiSeq. Using bsmap (Bisulphite Sequencing Mapping Platform) (Xi and Li 2009) we checked for bisulphite conversion efficiency (C→T in CH contexts) and quantified the percent methylation for each person at every CpG in the amplicons.

We validated differential methylation for 3 predicted DMRs: the loci in *NOS1*, *FAM134B*, and *OR2L13*, and tested one of the loci with unstable DMR prediction (at the *KCNQ5* gene). The loci tested were chosen on the basis of their potential functional relevance and technical ability to design bisulphite PCR primers covering CpGs that overlapped with array probes. **Supplemental Table S5** shows the primers used.

| Locus | Forward Primer | Reverse Primer |
|---------------------------|-------------------------------|---------------------------|
| chr12:117797179-117797517 | GGGGAAAAATTTATGTTTTAGAGAG | AAAATTCTTCCTCTACTCCCATAAC |
| chr6:73330181-73330482 | GGTTTTGTTGGTGATTAGGAGTAG | AAAAACAAAATAAACTTCCACCAC |
| chr1:248100298-248100643 | TTTTATTGTTTTTGGGGTTAATTAT | CACCAATATATAAAACAAAACCTTC |
| chr5:16508806-16509201 | TATTTTAATGTTGAATATTAGGAGGAAAA | AACCACTCCACCCTTAAATAAATAC |

Supplemental Table S5: Primers for bisulphite-converted DNA

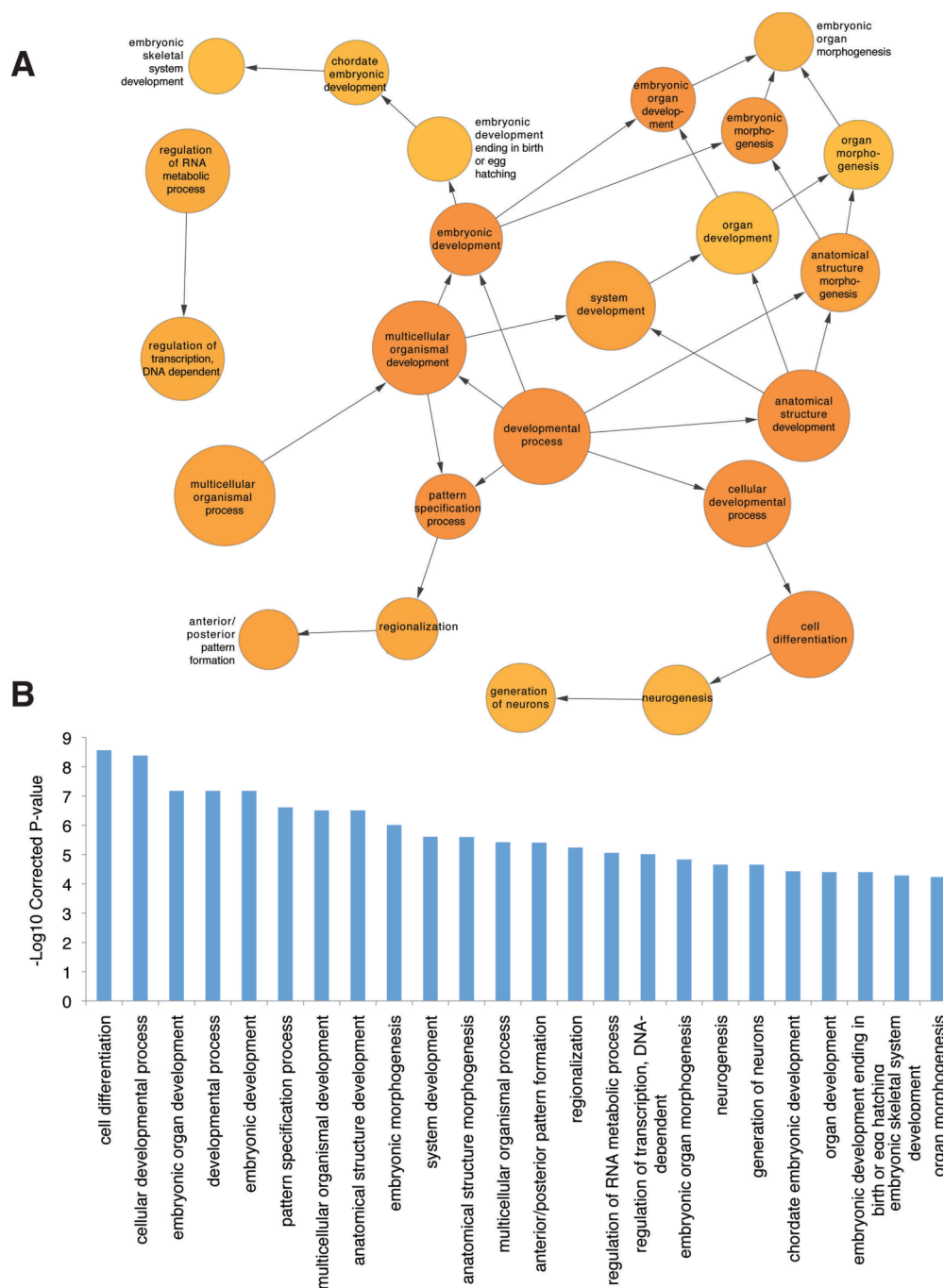
We showed a concordance of DNA methylation changes between microarray and sequencing-based approaches for all loci, but the degrees of difference of DNA methylation at the DMRs at *NOS1* and *KCNQ5* are insufficient to allow significant associations to be assigned statistically (**Supplemental Figure S6**).



Supplemental Figure S6: Results of massively-parallel bisulphite sequencing of the *NOS1* and *KCNQ5* DMRs. The p values were calculated using t tests combining all of the loci tested in the putative DMR and comparing between the ASD and TD groups.

Gene Ontology Enrichment Analysis

For age-associated DMRs obtained from bump-hunting, we plotted only the enrichment categories with a threshold of the p-value with a significance of 10^{-7} or lower. Enrichment category results are displayed in **Supplemental Figure S7** and the DMRs themselves in **Supplemental Table S6**.



Supplemental Figure S7: Gene ontology analysis of genes associated with age-related DMRs.

Panel A shows connectivity of the gene ontology categories significantly enriched for genes related to age-associated DMRs, plotted by corrected p-value in **Panel B**.

Supplemental Table S6: Age-associated candidate DMRs.

| chr | start | end | value | area | pns | indexStart | indexEnd | nprobes | avg | max | area.raw |
|-------|-------------|-------------|--------|-------|---------|------------|----------|---------|--------|--------|----------|
| chr2 | 176,986,460 | 176,987,465 | 0.029 | 0.318 | 132,157 | 265,348 | 265,358 | 11 | 0.732 | 0.855 | 8.054 |
| chr1 | 231,155,632 | 231,156,204 | -0.039 | 0.467 | 21,845 | 42,875 | 42,886 | 12 | -0.609 | -0.707 | 7.312 |
| chr6 | 32,118,295 | 32,118,457 | 0.026 | 0.290 | 189,023 | 380,814 | 380,824 | 11 | 0.659 | 0.753 | 7.250 |
| chr19 | 58,220,295 | 58,220,837 | 0.027 | 0.271 | 119,212 | 241,574 | 241,583 | 10 | 0.697 | 0.817 | 6.965 |
| chr14 | 24,641,021 | 24,641,852 | 0.048 | 0.529 | 67,268 | 134,784 | 134,794 | 11 | 0.606 | 0.635 | 6.670 |
| chr8 | 23,563,970 | 23,564,717 | 0.027 | 0.246 | 215,946 | 436,377 | 436,385 | 9 | 0.729 | 0.859 | 6.557 |
| chr1 | 110,610,899 | 110,612,044 | 0.033 | 0.267 | 12,843 | 25,512 | 25,519 | 8 | 0.799 | 0.850 | 6.392 |
| chr19 | 9,473,565 | 9,473,781 | 0.032 | 0.285 | 111,176 | 223,482 | 223,490 | 9 | 0.691 | 0.797 | 6.217 |
| chr16 | 66,612,955 | 66,613,334 | 0.037 | 0.334 | 88,644 | 175,913 | 175,921 | 9 | 0.663 | 0.794 | 5.970 |
| chr10 | 22,634,038 | 22,634,226 | 0.046 | 0.320 | 25,480 | 49,991 | 49,997 | 7 | 0.832 | 0.864 | 5.825 |
| chr13 | 79,169,714 | 79,170,303 | 0.027 | 0.217 | 63,849 | 127,718 | 127,725 | 8 | 0.718 | 0.788 | 5.745 |
| chr6 | 32,078,398 | 32,078,624 | -0.028 | 0.225 | 189,003 | 380,630 | 380,637 | 8 | -0.691 | -0.752 | 5.525 |
| chr2 | 177,029,459 | 177,030,171 | 0.027 | 0.192 | 132,199 | 265,502 | 265,508 | 7 | 0.775 | 0.798 | 5.425 |
| chr14 | 29,234,890 | 29,235,196 | 0.029 | 0.229 | 67,445 | 135,202 | 135,209 | 8 | 0.666 | 0.827 | 5.332 |
| chr5 | 87,441,081 | 87,441,969 | 0.031 | 0.249 | 177,623 | 353,048 | 353,055 | 8 | 0.659 | 0.812 | 5.272 |
| chr3 | 147,126,638 | 147,127,097 | 0.035 | 0.244 | 158,563 | 316,905 | 316,911 | 7 | 0.752 | 0.852 | 5.264 |
| chr7 | 27,205,200 | 27,205,658 | 0.025 | 0.223 | 202,198 | 409,826 | 409,834 | 9 | 0.583 | 0.696 | 5.251 |
| chr4 | 155,661,691 | 155,662,795 | 0.029 | 0.259 | 170,611 | 339,758 | 339,766 | 9 | 0.556 | 0.630 | 5.004 |
| chr11 | 86,383,182 | 86,383,430 | 0.041 | 0.286 | 44,708 | 90,004 | 90,010 | 7 | 0.707 | 0.738 | 4.950 |
| chr22 | 24,890,690 | 24,890,833 | 0.043 | 0.301 | 145,700 | 292,479 | 292,485 | 7 | 0.678 | 0.766 | 4.744 |

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|--------------|-------------|-------------|--------|-------|---------|---------|---------|---|--------|--------|-------|
| chr8 | 143,858,414 | 143,858,636 | 0.036 | 0.255 | 223,217 | 449,399 | 449,405 | 7 | 0.669 | 0.736 | 4.680 |
| chr7 | 99,775,422 | 99,775,558 | 0.034 | 0.240 | 207,428 | 419,896 | 419,902 | 7 | 0.666 | 0.745 | 4.661 |
| chr14 | 57,275,967 | 57,276,789 | 0.034 | 0.204 | 68,821 | 137,800 | 137,805 | 6 | 0.773 | 0.871 | 4.638 |
| chr7 | 27,225,058 | 27,225,143 | 0.029 | 0.172 | 202,219 | 409,908 | 409,913 | 6 | 0.772 | 0.848 | 4.631 |
| chr17 | 7,832,680 | 7,833,237 | 0.028 | 0.255 | 94,610 | 188,539 | 188,547 | 9 | 0.511 | 0.575 | 4.602 |
| chr1 | 248,020,436 | 248,020,812 | 0.043 | 0.258 | 23,207 | 45,341 | 45,346 | 6 | 0.749 | 0.833 | 4.492 |
| chr7 | 8,481,994 | 8,482,614 | 0.037 | 0.225 | 201,229 | 407,774 | 407,779 | 6 | 0.747 | 0.836 | 4.481 |
| chr16 | 87,864,324 | 87,865,062 | -0.050 | 0.300 | 91,530 | 181,608 | 181,613 | 6 | -0.721 | -0.750 | 4.325 |
| chr1 | 92,952,440 | 92,952,533 | 0.034 | 0.202 | 11,997 | 23,805 | 23,810 | 6 | 0.715 | 0.761 | 4.290 |
| chr2 | 74,875,227 | 74,875,387 | 0.042 | 0.254 | 125,794 | 254,086 | 254,091 | 6 | 0.713 | 0.788 | 4.276 |
| chr8 | 24,772,137 | 24,772,350 | 0.039 | 0.232 | 216,010 | 436,484 | 436,489 | 6 | 0.706 | 0.777 | 4.233 |
| chr14 | 54,413,218 | 54,413,931 | 0.052 | 0.310 | 68,535 | 137,302 | 137,307 | 6 | 0.704 | 0.806 | 4.221 |
| chr10 | 103,603,292 | 103,603,869 | 0.029 | 0.205 | 31,479 | 61,239 | 61,245 | 7 | 0.602 | 0.702 | 4.215 |
| chr1 | 91,301,204 | 91,301,962 | 0.029 | 0.172 | 11,874 | 23,499 | 23,504 | 6 | 0.695 | 0.766 | 4.170 |
| chr3 | 120,626,881 | 120,627,088 | 0.031 | 0.155 | 156,324 | 312,596 | 312,600 | 5 | 0.830 | 0.849 | 4.148 |
| chr2 | 10,182,878 | 10,183,227 | 0.043 | 0.258 | 120,711 | 244,696 | 244,701 | 6 | 0.690 | 0.742 | 4.140 |
| chr12 | 106,533,667 | 106,533,903 | 0.040 | 0.241 | 56,479 | 112,724 | 112,729 | 6 | 0.687 | 0.745 | 4.119 |
| chr11 | 2,292,751 | 2,292,914 | 0.036 | 0.253 | 36,185 | 72,072 | 72,078 | 7 | 0.588 | 0.751 | 4.116 |
| chr6 | 100,903,561 | 100,903,909 | 0.025 | 0.148 | 193,518 | 391,945 | 391,950 | 6 | 0.686 | 0.807 | 4.113 |
| chr6 | 85,474,028 | 85,474,209 | 0.030 | 0.178 | 192,887 | 390,662 | 390,667 | 6 | 0.680 | 0.859 | 4.083 |
| chr11 | 94,278,324 | 94,278,603 | 0.032 | 0.259 | 45,079 | 90,675 | 90,682 | 8 | 0.508 | 0.574 | 4.061 |
| chr5 | 87,980,882 | 87,981,253 | 0.026 | 0.158 | 177,666 | 353,160 | 353,165 | 6 | 0.670 | 0.802 | 4.020 |
| chr2 | 220,299,643 | 220,299,900 | 0.032 | 0.190 | 134,869 | 270,356 | 270,361 | 6 | 0.667 | 0.762 | 4.003 |

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|--------------|-------------|-------------|--------|-------|---------|---------|---------|---|--------|--------|-------|
| chr22 | 30,476,089 | 30,476,285 | 0.033 | 0.230 | 146,109 | 293,345 | 293,351 | 7 | 0.565 | 0.685 | 3.953 |
| chr5 | 78,407,552 | 78,407,683 | 0.029 | 0.171 | 177,238 | 352,385 | 352,390 | 6 | 0.657 | 0.800 | 3.943 |
| chr6 | 29,943,268 | 29,943,480 | 0.038 | 0.308 | 188,133 | 374,577 | 374,584 | 8 | 0.489 | 0.583 | 3.911 |
| chr1 | 79,472,282 | 79,472,452 | 0.030 | 0.152 | 11,278 | 22,419 | 22,423 | 5 | 0.779 | 0.847 | 3.895 |
| chr18 | 44,526,430 | 44,527,026 | 0.031 | 0.246 | 106,733 | 214,413 | 214,420 | 8 | 0.485 | 0.657 | 3.883 |
| chr13 | 100,624,279 | 100,624,373 | 0.026 | 0.128 | 64,614 | 129,099 | 129,103 | 5 | 0.776 | 0.832 | 3.880 |
| chr5 | 43,017,982 | 43,018,629 | 0.045 | 0.227 | 175,429 | 349,043 | 349,047 | 5 | 0.760 | 0.817 | 3.802 |
| chr4 | 85,414,016 | 85,414,486 | 0.027 | 0.134 | 167,599 | 334,300 | 334,304 | 5 | 0.754 | 0.792 | 3.772 |
| chr22 | 24,181,191 | 24,181,270 | 0.034 | 0.169 | 145,604 | 292,299 | 292,303 | 5 | 0.754 | 0.818 | 3.771 |
| chr5 | 172,672,390 | 172,672,817 | 0.027 | 0.136 | 183,336 | 363,882 | 363,886 | 5 | 0.749 | 0.773 | 3.744 |
| chr1 | 151,810,586 | 151,810,904 | 0.031 | 0.187 | 14,673 | 29,289 | 29,294 | 6 | 0.621 | 0.691 | 3.729 |
| chr3 | 147,127,579 | 147,128,157 | 0.034 | 0.169 | 158,564 | 316,915 | 316,919 | 5 | 0.744 | 0.861 | 3.722 |
| chr7 | 100,463,416 | 100,464,145 | -0.059 | 0.353 | 207,617 | 420,346 | 420,351 | 6 | -0.615 | -0.639 | 3.692 |
| chr13 | 79,170,627 | 79,171,230 | 0.028 | 0.139 | 63,849 | 127,727 | 127,731 | 5 | 0.737 | 0.838 | 3.683 |
| chr4 | 85,402,870 | 85,403,409 | 0.027 | 0.137 | 167,592 | 334,286 | 334,290 | 5 | 0.732 | 0.761 | 3.658 |
| chr16 | 86,547,203 | 86,547,544 | 0.027 | 0.137 | 91,139 | 180,813 | 180,817 | 5 | 0.731 | 0.767 | 3.654 |
| chr11 | 86,085,623 | 86,086,005 | 0.026 | 0.158 | 44,685 | 89,972 | 89,977 | 6 | 0.609 | 0.731 | 3.652 |
| chr7 | 19,146,032 | 19,146,555 | 0.025 | 0.123 | 201,554 | 408,365 | 408,369 | 5 | 0.730 | 0.799 | 3.650 |
| chr2 | 223,164,831 | 223,164,925 | 0.027 | 0.134 | 135,087 | 270,748 | 270,752 | 5 | 0.727 | 0.805 | 3.637 |
| chr5 | 176,827,082 | 176,827,793 | 0.040 | 0.237 | 183,904 | 364,948 | 364,953 | 6 | 0.599 | 0.723 | 3.597 |
| chr3 | 147,125,712 | 147,125,782 | 0.031 | 0.157 | 158,563 | 316,888 | 316,892 | 5 | 0.717 | 0.828 | 3.586 |
| chr6 | 85,482,570 | 85,483,055 | 0.032 | 0.159 | 192,892 | 390,680 | 390,684 | 5 | 0.715 | 0.793 | 3.573 |
| chr6 | 11,044,877 | 11,044,974 | 0.067 | 0.269 | 186,057 | 369,233 | 369,236 | 4 | 0.892 | 0.905 | 3.568 |

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|--------------|-------------|-------------|--------|-------|---------|---------|---------|---|--------|--------|-------|
| chr2 | 176,948,693 | 176,948,759 | 0.029 | 0.147 | 132,115 | 265,209 | 265,213 | 5 | 0.706 | 0.784 | 3.528 |
| chr1 | 91,190,366 | 91,190,891 | 0.028 | 0.139 | 11,865 | 23,457 | 23,461 | 5 | 0.705 | 0.795 | 3.527 |
| chr17 | 46,685,292 | 46,685,448 | 0.028 | 0.141 | 99,899 | 199,582 | 199,586 | 5 | 0.701 | 0.744 | 3.505 |
| chr16 | 1,593,152 | 1,593,766 | -0.034 | 0.202 | 83,002 | 165,085 | 165,090 | 6 | -0.583 | -0.705 | 3.499 |
| chr16 | 68,482,591 | 68,482,821 | 0.030 | 0.148 | 89,083 | 176,948 | 176,952 | 5 | 0.697 | 0.806 | 3.483 |
| chr11 | 2,891,065 | 2,891,118 | 0.031 | 0.154 | 36,402 | 72,730 | 72,734 | 5 | 0.691 | 0.737 | 3.456 |
| chr16 | 51,187,388 | 51,187,807 | 0.030 | 0.148 | 87,625 | 174,051 | 174,055 | 5 | 0.686 | 0.801 | 3.429 |
| chr10 | 93,805,441 | 93,805,870 | 0.028 | 0.139 | 30,293 | 58,645 | 58,649 | 5 | 0.669 | 0.772 | 3.344 |
| chr17 | 78,999,347 | 78,999,895 | -0.032 | 0.162 | 104,247 | 208,702 | 208,706 | 5 | -0.668 | -0.739 | 3.342 |
| chr14 | 100,069,535 | 100,069,840 | 0.055 | 0.222 | 72,269 | 144,115 | 144,118 | 4 | 0.827 | 0.851 | 3.307 |
| chr22 | 40,417,285 | 40,417,869 | -0.043 | 0.214 | 147,204 | 295,668 | 295,672 | 5 | -0.653 | -0.769 | 3.264 |
| chr12 | 113,916,473 | 113,916,664 | 0.030 | 0.122 | 57,370 | 114,617 | 114,620 | 4 | 0.811 | 0.828 | 3.242 |
| chr8 | 97,157,756 | 97,158,052 | 0.026 | 0.128 | 220,041 | 443,885 | 443,889 | 5 | 0.648 | 0.741 | 3.240 |
| chr1 | 164,545,553 | 164,546,143 | 0.035 | 0.174 | 16,595 | 33,260 | 33,264 | 5 | 0.646 | 0.810 | 3.229 |
| chr3 | 14,614,882 | 14,615,579 | -0.032 | 0.161 | 149,961 | 301,002 | 301,006 | 5 | -0.641 | -0.692 | 3.203 |
| chr6 | 101,846,779 | 101,846,872 | 0.026 | 0.130 | 193,554 | 392,047 | 392,051 | 5 | 0.638 | 0.692 | 3.191 |
| chr2 | 176,964,506 | 176,964,720 | 0.030 | 0.118 | 132,130 | 265,271 | 265,274 | 4 | 0.798 | 0.830 | 3.190 |
| chr6 | 73,329,988 | 73,330,358 | 0.036 | 0.217 | 192,434 | 389,727 | 389,732 | 6 | 0.529 | 0.610 | 3.176 |
| chr1 | 119,535,693 | 119,535,986 | 0.038 | 0.153 | 13,653 | 27,185 | 27,188 | 4 | 0.794 | 0.872 | 3.175 |
| chr11 | 123,066,529 | 123,067,275 | 0.030 | 0.181 | 47,241 | 94,847 | 94,852 | 6 | 0.529 | 0.683 | 3.174 |
| chr1 | 228,400,217 | 228,400,693 | 0.034 | 0.136 | 21,459 | 42,125 | 42,128 | 4 | 0.788 | 0.822 | 3.153 |
| chr8 | 56,015,399 | 56,015,785 | 0.026 | 0.105 | 218,073 | 440,284 | 440,287 | 4 | 0.784 | 0.808 | 3.135 |
| chr5 | 170,288,742 | 170,289,070 | 0.034 | 0.171 | 183,006 | 363,218 | 363,222 | 5 | 0.625 | 0.864 | 3.127 |

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|--------------|-------------|-------------|--------|-------|---------|---------|---------|---|--------|--------|-------|
| chr8 | 25,898,191 | 25,898,539 | 0.031 | 0.126 | 216,106 | 436,652 | 436,655 | 4 | 0.778 | 0.803 | 3.111 |
| chr5 | 126,626,348 | 126,626,364 | 0.025 | 0.099 | 179,378 | 356,130 | 356,133 | 4 | 0.777 | 0.826 | 3.106 |
| chr11 | 134,147,143 | 134,147,634 | 0.034 | 0.136 | 48,559 | 97,232 | 97,235 | 4 | 0.773 | 0.855 | 3.091 |
| chr19 | 17,958,339 | 17,958,736 | 0.032 | 0.159 | 113,027 | 227,806 | 227,810 | 5 | 0.618 | 0.744 | 3.091 |
| chr19 | 57,182,844 | 57,183,268 | 0.031 | 0.124 | 119,037 | 241,073 | 241,076 | 4 | 0.768 | 0.814 | 3.070 |
| chr10 | 28,035,631 | 28,035,894 | 0.038 | 0.267 | 25,811 | 50,667 | 50,673 | 7 | 0.439 | 0.487 | 3.070 |
| chr13 | 79,177,877 | 79,177,925 | 0.027 | 0.107 | 63,854 | 127,748 | 127,751 | 4 | 0.759 | 0.790 | 3.037 |
| chr7 | 101,005,910 | 101,006,089 | 0.031 | 0.215 | 207,774 | 420,718 | 420,724 | 7 | 0.433 | 0.521 | 3.032 |
| chr11 | 64,146,487 | 64,146,822 | -0.034 | 0.172 | 41,583 | 83,185 | 83,189 | 5 | -0.606 | -0.642 | 3.030 |
| chr7 | 153,584,416 | 153,584,609 | 0.034 | 0.136 | 211,900 | 428,260 | 428,263 | 4 | 0.757 | 0.787 | 3.029 |
| chr1 | 75,595,919 | 75,596,336 | 0.029 | 0.118 | 11,085 | 22,047 | 22,050 | 4 | 0.757 | 0.834 | 3.029 |
| chr8 | 11,555,178 | 11,555,548 | 0.029 | 0.114 | 214,859 | 434,416 | 434,419 | 4 | 0.756 | 0.799 | 3.025 |
| chr12 | 54,071,090 | 54,071,194 | 0.033 | 0.163 | 53,207 | 106,359 | 106,363 | 5 | 0.605 | 0.704 | 3.024 |
| chr1 | 39,957,387 | 39,957,400 | 0.026 | 0.105 | 8,008 | 16,050 | 16,053 | 4 | 0.752 | 0.768 | 3.009 |
| chr5 | 87,968,528 | 87,968,749 | 0.029 | 0.115 | 177,652 | 353,113 | 353,116 | 4 | 0.752 | 0.830 | 3.008 |
| chr10 | 88,149,210 | 88,149,632 | 0.040 | 0.159 | 29,828 | 57,728 | 57,731 | 4 | 0.752 | 0.813 | 3.007 |
| chr1 | 18,959,268 | 18,959,625 | 0.025 | 0.101 | 4,757 | 9,510 | 9,513 | 4 | 0.742 | 0.756 | 2.966 |
| chr2 | 239,139,911 | 239,140,190 | 0.032 | 0.195 | 136,760 | 273,781 | 273,786 | 6 | 0.494 | 0.662 | 2.962 |
| chr16 | 29,625,216 | 29,625,259 | 0.036 | 0.145 | 86,317 | 171,434 | 171,437 | 4 | 0.739 | 0.794 | 2.958 |
| chr3 | 147,111,120 | 147,111,308 | 0.024 | 0.098 | 158,547 | 316,846 | 316,849 | 4 | 0.737 | 0.803 | 2.947 |
| chr11 | 35,441,558 | 35,441,900 | 0.029 | 0.144 | 39,114 | 78,143 | 78,147 | 5 | 0.588 | 0.723 | 2.942 |
| chr2 | 200,329,654 | 200,329,680 | 0.028 | 0.113 | 133,200 | 267,328 | 267,331 | 4 | 0.735 | 0.837 | 2.941 |
| chr5 | 132,083,532 | 132,084,068 | 0.032 | 0.127 | 179,674 | 356,716 | 356,719 | 4 | 0.734 | 0.835 | 2.938 |

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|-------|-------------|-------------|--------|-------|---------|---------|---------|---|--------|--------|-------|
| | | | | | | | | | | | 2.928 |
| chr8 | 25,905,478 | 25,905,811 | 0.028 | 0.112 | 216,114 | 436,684 | 436,687 | 4 | 0.732 | 0.775 | |
| | | | | | 202,239 | 409,982 | | | | | |
| chr7 | 27,245,018 | 27,245,747 | 0.026 | 0.103 | | | 409,985 | 4 | 0.724 | 0.765 | 2.896 |
| chr1 | 154,475,068 | 154,475,269 | 0.032 | 0.127 | 15,125 | 30,199 | 30,202 | 4 | 0.723 | 0.793 | 2.891 |
| chr18 | 70,534,298 | 70,535,005 | 0.027 | 0.135 | 107,430 | 215,904 | 215,908 | 5 | 0.578 | 0.744 | 2.888 |
| chr5 | 87,979,441 | 87,979,871 | 0.026 | 0.105 | 177,664 | 353,155 | 353,158 | 4 | 0.721 | 0.816 | 2.884 |
| chr14 | 95,239,381 | 95,239,751 | 0.026 | 0.131 | 71,852 | 143,386 | 143,390 | 5 | 0.576 | 0.861 | 2.882 |
| chr11 | 66,102,055 | 66,102,352 | -0.029 | 0.144 | 42,230 | 84,801 | 84,805 | 5 | -0.572 | -0.642 | 2.861 |
| | | | | | 193,460 | 391,824 | | | | | |
| chr6 | 100,054,585 | 100,054,817 | 0.026 | 0.104 | | | 391,827 | 4 | 0.714 | 0.855 | 2.855 |
| | | | | 0.199 | 109,187 | | | | | | |
| chr19 | 2,046,085 | 2,046,350 | -0.050 | | | 219,324 | 219,327 | 4 | -0.711 | -0.744 | 2.846 |
| chr6 | 100,442,105 | 100,442,151 | 0.037 | 0.150 | 193,479 | 391,871 | 391,874 | 4 | 0.710 | 0.720 | 2.841 |
| chr18 | 49,868,378 | 49,868,552 | 0.027 | 0.108 | 106,986 | 214,941 | 214,944 | 4 | 0.708 | 0.758 | 2.832 |
| chr3 | 62,354,991 | 62,355,443 | 0.030 | 0.118 | 153,866 | 308,533 | 308,536 | 4 | 0.708 | 0.780 | 2.832 |
| chr4 | 174,430,487 | 174,431,058 | 0.024 | 0.096 | 171,282 | 340,983 | 340,986 | 4 | 0.706 | 0.731 | 2.825 |
| chr3 | 187,387,555 | 187,387,734 | 0.031 | 0.124 | 160,929 | 321,238 | 321,241 | 4 | 0.706 | 0.841 | 2.823 |
| chr10 | 22,634,578 | 22,635,028 | 0.025 | 0.102 | 25,480 | 50,000 | 50,003 | 4 | 0.699 | 0.782 | 2.796 |
| chr6 | 134,213,992 | | | | | | | | | | |
| | | 134,214,307 | 0.027 | 0.109 | 195,293 | 395,191 | 395,194 | 4 | 0.699 | 0.753 | 2.794 |
| | | 40,715,281 | | | | | | | | | |
| chr17 | 40,715,222 | | -0.026 | 0.132 | 98,728 | 196,894 | 196,898 | 5 | -0.558 | -0.644 | 2.788 |
| chr4 | 52,942,997 | 52,943,247 | 0.042 | 0.169 | 166,053 | 331,543 | 331,546 | 4 | 0.696 | 0.757 | 2.784 |
| chr2 | 223,161,771 | 223,162,128 | 0.032 | 0.127 | 135,085 | 270,727 | 270,730 | 4 | 0.695 | 0.792 | 2.780 |
| chr11 | 111,385,450 | 111,385,659 | 0.027 | 0.109 | 45,879 | 92,155 | 92,158 | 4 | 0.695 | 0.805 | 2.779 |
| chr15 | 60,288,082 | 60,288,404 | 0.026 | 0.103 | 77,285 | 154,102 | 154,105 | 4 | 0.694 | 0.759 | 2.777 |
| | | | | | | | 287,544 | | | | |
| chr21 | 36,041,605 | 36,041,699 | 0.033 | 0.134 | 143,100 | 287,541 | | 4 | 0.694 | 0.716 | 2.775 |
| | | | | | | 397,938 | | | | | |
| chr6 | 156,718,177 | 156,718,546 | 0.039 | 0.193 | 196,828 | | 397,942 | 5 | 0.554 | 0.626 | 2.770 |

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|--------------|-------------|-------------|--------|-------|---------|---------|---------|---|--------|--------|-------|
| chr8 | 98,290,310 | 98,290,372 | 0.034 | 0.137 | 220,135 | 444,065 | 444,068 | 4 | 0.692 | 0.826 | 2.767 |
| chr15 | 58,357,922 | 58,357,989 | 0.028 | 0.110 | 77,129 | 153,822 | 153,825 | 4 | 0.691 | 0.723 | 2.764 |
| chr4 | 48,486,087 | 48,486,472 | 0.026 | 0.106 | 165,951 | 331,335 | 331,338 | 4 | 0.687 | 0.752 | 2.748 |
| chr17 | 36,719,518 | 36,719,937 | 0.034 | 0.172 | 97,771 | 194,856 | 194,860 | 5 | 0.549 | 0.726 | 2.744 |
| chr1 | 19,665,070 | 19,665,240 | 0.040 | 0.161 | 4,926 | 9,817 | 9,820 | 4 | 0.686 | 0.731 | 2.744 |
| chr10 | 126,136,228 | 126,136,709 | 0.059 | 0.237 | 33,326 | 64,892 | 64,895 | 4 | 0.683 | 0.755 | 2.733 |
| chr10 | 50,604,330 | 50,604,569 | 0.024 | 0.097 | 27,342 | 53,347 | 53,350 | 4 | 0.680 | 0.779 | 2.721 |
| chr16 | 2,563,274 | 2,563,560 | 0.024 | 0.097 | 83,480 | 166,190 | 166,193 | 4 | 0.679 | 0.803 | 2.717 |
| chr19 | 10,397,612 | 10,397,780 | 0.031 | 0.125 | 111,331 | 223,899 | 223,902 | 4 | 0.678 | 0.766 | 2.713 |
| chr13 | 37,004,721 | 37,004,812 | 0.025 | 0.127 | 61,968 | 124,283 | 124,287 | 5 | 0.542 | 0.570 | 2.711 |
| chr17 | 61,778,366 | 61,778,813 | 0.025 | 0.100 | 101,437 | 202,761 | 202,764 | 4 | 0.675 | 0.730 | 2.701 |
| chr17 | 58,216,297 | 58,216,651 | 0.030 | 0.119 | 101,114 | 202,101 | 202,104 | 4 | 0.672 | 0.755 | 2.688 |
| chr20 | 55,964,998 | 55,965,497 | 0.035 | 0.138 | 141,209 | 283,686 | 283,689 | 4 | 0.671 | 0.781 | 2.682 |
| chr2 | 157,176,971 | 157,177,345 | 0.035 | 0.141 | 130,820 | 262,911 | 262,914 | 4 | 0.671 | 0.825 | 2.682 |
| chr8 | 143,545,478 | 143,545,949 | 0.034 | 0.170 | 223,102 | 449,175 | 449,179 | 5 | 0.536 | 0.619 | 2.680 |
| chr8 | 1,273,592 | 1,273,856 | 0.028 | 0.139 | 213,656 | 432,090 | 432,094 | 5 | 0.535 | 0.646 | 2.674 |
| chr12 | 66,627,900 | 66,628,232 | -0.025 | 0.099 | 54,519 | 109,239 | 109,242 | 4 | -0.667 | -0.715 | 2.668 |
| chr14 | 29,243,404 | 29,243,690 | 0.028 | 0.114 | 67,454 | 135,237 | 135,240 | 4 | 0.666 | 0.790 | 2.665 |
| chr1 | 41,119,634 | 41,119,988 | 0.030 | 0.121 | 8,190 | 16,458 | 16,461 | 4 | 0.660 | 0.741 | 2.638 |
| chr6 | 30,653,512 | 30,653,659 | -0.028 | 0.112 | 188,431 | 376,470 | 376,473 | 4 | -0.659 | -0.679 | 2.635 |
| chr4 | 151,500,631 | 151,501,298 | 0.037 | 0.148 | 170,283 | 339,171 | 339,174 | 4 | 0.659 | 0.774 | 2.634 |
| chr16 | 85,932,591 | 85,932,853 | 0.031 | 0.123 | 90,989 | 180,519 | 180,522 | 4 | 0.658 | 0.752 | 2.632 |
| chr2 | 177,053,274 | 177,053,292 | 0.028 | 0.113 | 132,211 | 265,539 | 265,542 | 4 | 0.658 | 0.724 | 2.631 |

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|-------|-------------|-------------|--------|-------|---------|---------|---------|---|--------|--------|-------|
| | 24,844,846 | | | | | | | | | | |
| chr13 | | 24,844,938 | 0.028 | 0.168 | 60,981 | 122,394 | 122,399 | 6 | 0.438 | 0.541 | 2.631 |
| chr12 | 56,414,442 | 56,414,533 | 0.025 | 0.099 | 53,637 | 107,388 | 107,391 | 4 | 0.654 | 0.701 | 2.616 |
| chr11 | 46,383,031 | 46,383,209 | 0.028 | 0.111 | 39,798 | 79,438 | 79,441 | 4 | 0.653 | 0.724 | 2.611 |
| chr19 | 18,260,330 | 18,260,515 | -0.033 | 0.131 | 113,106 | 227,997 | 228,000 | 4 | -0.650 | -0.685 | 2.599 |
| chr12 | 126,675,667 | 126,676,048 | 0.026 | 0.103 | 59,051 | 118,197 | 118,200 | 4 | 0.650 | 0.770 | 2.598 |
| chr6 | 110,736,772 | 110,737,053 | -0.033 | 0.163 | 194,095 | 393,045 | 393,049 | 5 | -0.517 | -0.584 | 2.586 |
| chr15 | 67,356,310 | 67,356,942 | 0.035 | 0.174 | 78,156 | 155,651 | 155,655 | 5 | 0.516 | 0.559 | 2.579 |
| chr8 | 61,777,711 | 61,778,137 | -0.036 | 0.146 | 218,407 | 440,878 | 440,881 | 4 | -0.643 | -0.724 | 2.572 |
| chr1 | 6,515,580 | 6,515,748 | 0.036 | 0.145 | 2,387 | 5,290 | 5,293 | 4 | 0.643 | 0.703 | 2.570 |
| chr11 | 17,803,160 | 17,803,421 | -0.036 | 0.145 | 38,057 | 76,022 | 76,025 | 4 | -0.642 | -0.684 | 2.567 |
| chr17 | 46,618,919 | 46,619,555 | 0.026 | 0.104 | 99,835 | 199,378 | 199,381 | 4 | 0.641 | 0.826 | 2.565 |
| chr10 | 99,734,416 | 99,734,912 | 0.034 | 0.169 | 30,914 | 59,888 | 59,892 | 5 | 0.513 | 0.616 | 2.564 |
| chr4 | 175,132,842 | 175,133,151 | 0.024 | 0.096 | 171,327 | 341,091 | 341,094 | 4 | 0.640 | 0.744 | 2.561 |
| chr1 | 200,009,830 | 200,010,283 | 0.024 | 0.097 | 18,525 | 36,715 | 36,718 | 4 | 0.638 | 0.685 | 2.551 |
| chr5 | 54,518,745 | 54,519,159 | 0.028 | 0.113 | 175,795 | 349,774 | 349,777 | 4 | 0.637 | 0.754 | 2.549 |
| chr6 | 30,070,059 | 30,070,403 | 0.038 | 0.339 | 188,216 | 375,015 | 375,023 | 9 | 0.283 | 0.533 | 2.543 |
| chr20 | 33,762,474 | 33,762,943 | 0.028 | 0.111 | 139,752 | 280,319 | 280,322 | 4 | 0.635 | 0.727 | 2.540 |
| chr7 | 94,284,865 | 94,284,900 | 0.030 | 0.121 | 206,839 | 418,369 | 418,372 | 4 | 0.634 | 0.835 | 2.535 |
| chr2 | 175,208,588 | 175,208,761 | 0.026 | 0.103 | 131,985 | 264,936 | 264,939 | 4 | 0.631 | 0.743 | 2.526 |
| chr14 | 94,392,718 | 94,392,932 | -0.030 | 0.121 | 71,715 | 143,118 | 143,121 | 4 | -0.630 | -0.671 | 2.519 |
| chr12 | 50,426,531 | 50,427,095 | 0.032 | 0.127 | 52,527 | 104,971 | 104,974 | 4 | 0.628 | 0.698 | 2.512 |
| chr1 | 17,085,860 | 17,086,071 | 0.030 | 0.121 | 4,431 | 8,941 | 8,944 | 4 | 0.627 | 0.684 | 2.508 |
| chr17 | 27,038,861 | 27,039,058 | 0.028 | 0.110 | 96,631 | 192,556 | 192,559 | 4 | 0.627 | 0.717 | 2.506 |

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|--------------|-------------|-------------|--------|-------|---------|---------|---------|---|--------|--------|-------|
| chr12 | 11,653,278 | 11,653,827 | 0.024 | 0.098 | 50,245 | 100,630 | 100,633 | 4 | 0.626 | 0.842 | 2.505 |
| chr21 | 36,399,146 | 36,399,540 | 0.041 | 0.166 | 143,128 | 287,597 | 287,600 | 4 | 0.625 | 0.684 | 2.498 |
| chr7 | 142,494,148 | 142,494,244 | 0.028 | 0.113 | 210,754 | 426,012 | 426,015 | 4 | 0.622 | 0.724 | 2.490 |
| chr2 | 74,663,416 | 74,663,698 | 0.026 | 0.104 | 125,719 | 253,871 | 253,874 | 4 | 0.622 | 0.725 | 2.487 |
| chr1 | 91,300,288 | 91,300,446 | 0.025 | 0.102 | 11,873 | 23,494 | 23,497 | 4 | 0.622 | 0.653 | 2.486 |
| chr17 | 42,733,527 | 42,733,600 | 0.033 | 0.130 | 99,202 | 198,076 | 198,079 | 4 | 0.611 | 0.835 | 2.443 |
| chr10 | 110,225,900 | 110,226,387 | 0.025 | 0.099 | 31,992 | 62,298 | 62,301 | 4 | 0.611 | 0.627 | 2.443 |
| chr1 | 3,473,665 | 3,474,376 | -0.028 | 0.111 | 1,736 | 3,933 | 3,936 | 4 | -0.610 | -0.781 | 2.440 |
| chr13 | 28,545,214 | 28,545,566 | 0.025 | 0.098 | 61,363 | 123,182 | 123,185 | 4 | 0.607 | 0.697 | 2.429 |
| chr3 | 194,408,516 | 194,408,901 | 0.036 | 0.143 | 161,474 | 322,164 | 322,167 | 4 | 0.607 | 0.695 | 2.427 |
| chr1 | 33,231,272 | 33,231,382 | 0.035 | 0.140 | 7,123 | 14,255 | 14,258 | 4 | 0.603 | 0.623 | 2.412 |
| chr17 | 1,881,005 | 1,881,333 | 0.036 | 0.145 | 93,375 | 185,564 | 185,567 | 4 | 0.601 | 0.611 | 2.404 |
| chr1 | 115,881,130 | 115,881,259 | 0.029 | 0.114 | 13,375 | 26,633 | 26,636 | 4 | 0.601 | 0.698 | 2.403 |
| chr4 | 109,093,158 | 109,093,243 | 0.025 | 0.101 | 168,519 | 336,010 | 336,013 | 4 | 0.595 | 0.663 | 2.380 |
| chr2 | 186,603,398 | 186,603,639 | 0.030 | 0.120 | 132,626 | 266,293 | 266,296 | 4 | 0.592 | 0.691 | 2.369 |
| chr2 | 220,196,530 | 220,196,755 | 0.031 | 0.125 | 134,841 | 270,288 | 270,291 | 4 | 0.592 | 0.800 | 2.368 |
| chr7 | 101,512,529 | 101,513,100 | 0.025 | 0.099 | 207,835 | 420,826 | 420,829 | 4 | 0.592 | 0.650 | 2.367 |
| chr13 | 112,709,256 | 112,709,550 | 0.024 | 0.097 | 65,560 | 130,940 | 130,943 | 4 | 0.590 | 0.803 | 2.361 |
| chr10 | 94,451,351 | 94,451,736 | 0.026 | 0.105 | 30,344 | 58,759 | 58,762 | 4 | 0.589 | 0.644 | 2.358 |
| chr6 | 29,521,781 | 29,521,803 | 0.030 | 0.121 | 187,956 | 373,759 | 373,762 | 4 | 0.588 | 0.632 | 2.352 |
| chr17 | 4,648,566 | 4,648,949 | 0.034 | 0.135 | 93,932 | 186,796 | 186,799 | 4 | 0.586 | 0.755 | 2.346 |
| chr19 | 11,529,947 | 11,530,065 | 0.029 | 0.145 | 111,665 | 224,649 | 224,653 | 5 | 0.468 | 0.552 | 2.342 |
| chr8 | 10,261,972 | 10,262,221 | 0.029 | 0.117 | 214,638 | 433,984 | 433,987 | 4 | 0.585 | 0.697 | 2.338 |

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|--------------|-------------|-------------|--------|-------|---------|---------|---------|---|--------|--------|-------|
| chr11 | 125,036,088 | 125,036,420 | 0.030 | 0.119 | 47,523 | 95,377 | 95,380 | 4 | 0.584 | 0.668 | 2.334 |
| chr1 | 4,770,676 | 4,771,201 | 0.034 | 0.136 | 2,015 | 4,574 | 4,577 | 4 | 0.583 | 0.707 | 2.333 |
| chr12 | 58,003,774 | 58,003,965 | 0.025 | 0.127 | 54,003 | 108,274 | 108,278 | 5 | 0.466 | 0.629 | 2.331 |
| chr11 | 9,025,730 | 9,026,308 | 0.035 | 0.140 | 37,246 | 74,531 | 74,534 | 4 | 0.580 | 0.758 | 2.322 |
| chr11 | 334,298 | 334,833 | 0.035 | 0.138 | 35,235 | 69,532 | 69,535 | 4 | 0.576 | 0.582 | 2.305 |
| chr17 | 7,350,001 | 7,350,413 | 0.026 | 0.103 | 94,440 | 188,076 | 188,079 | 4 | 0.574 | 0.627 | 2.297 |
| chr1 | 47,882,686 | 47,883,234 | 0.025 | 0.102 | 9,213 | 18,622 | 18,625 | 4 | 0.574 | 0.790 | 2.295 |
| chr10 | 8,097,331 | 8,097,689 | 0.032 | 0.128 | 24,556 | 48,236 | 48,239 | 4 | 0.573 | 0.672 | 2.293 |
| chr4 | 41,646,293 | 41,646,672 | 0.026 | 0.105 | 165,708 | 330,788 | 330,791 | 4 | 0.572 | 0.657 | 2.290 |
| chr13 | 112,717,207 | 112,717,707 | 0.031 | 0.126 | 65,571 | 130,968 | 130,971 | 4 | 0.571 | 0.630 | 2.284 |
| chr1 | 228,246,632 | 228,247,135 | 0.027 | 0.108 | 21,403 | 41,970 | 41,973 | 4 | 0.570 | 0.623 | 2.282 |
| chr22 | 17,083,412 | 17,083,727 | 0.025 | 0.101 | 144,702 | 290,507 | 290,510 | 4 | 0.570 | 0.658 | 2.280 |
| chr8 | 145,106,246 | 145,106,582 | 0.039 | 0.155 | 223,787 | 450,753 | 450,756 | 4 | 0.568 | 0.584 | 2.272 |
| chr6 | 125,855,124 | 125,855,421 | 0.027 | 0.110 | 194,820 | 394,326 | 394,329 | 4 | 0.565 | 0.583 | 2.261 |
| chr6 | 170,597,326 | 170,597,588 | -0.036 | 0.145 | 198,758 | 401,914 | 401,917 | 4 | -0.562 | -0.663 | 2.248 |
| chr5 | 172,110,211 | 172,110,579 | 0.028 | 0.113 | 183,213 | 363,608 | 363,611 | 4 | 0.559 | 0.695 | 2.237 |
| chr11 | 1,483,731 | 1,483,973 | 0.026 | 0.103 | 35,852 | 71,128 | 71,131 | 4 | 0.559 | 0.663 | 2.236 |
| chr12 | 113,913,695 | 113,914,222 | 0.028 | 0.112 | 57,369 | 114,609 | 114,612 | 4 | 0.557 | 0.748 | 2.229 |
| chr1 | 236,557,182 | 236,557,682 | -0.032 | 0.127 | 22,345 | 43,778 | 43,781 | 4 | -0.557 | -0.739 | 2.229 |
| chr3 | 100,712,058 | 100,712,345 | -0.024 | 0.097 | 155,320 | 310,850 | 310,853 | 4 | -0.555 | -0.560 | 2.221 |
| chr15 | 45,996,521 | 45,996,787 | 0.028 | 0.112 | 76,437 | 152,529 | 152,532 | 4 | 0.553 | 0.665 | 2.213 |
| chr15 | 37,387,304 | 37,387,577 | 0.032 | 0.160 | 75,453 | 150,422 | 150,426 | 5 | 0.443 | 0.507 | 2.213 |
| chr22 | 19,748,777 | 19,749,188 | 0.032 | 0.126 | 145,030 | 291,203 | 291,206 | 4 | 0.552 | 0.710 | 2.208 |

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|-------|-------------|-------------|--------|-------|---------|---------|---------|----|--------|--------|-------|
| chr19 | 3,097,565 | 3,097,728 | -0.028 | 0.111 | 109,601 | 220,187 | 220,190 | 4 | -0.549 | -0.559 | 2.197 |
| chr16 | 66,400,320 | 66,400,411 | -0.032 | 0.126 | 88,601 | 175,821 | 175,824 | 4 | -0.549 | -0.699 | 2.196 |
| chr1 | 228,225,533 | 228,225,687 | 0.030 | 0.119 | 21,398 | 41,960 | 41,963 | 4 | 0.549 | 0.571 | 2.194 |
| chr19 | 15,530,606 | 15,530,870 | -0.025 | 0.100 | 112,528 | 226,742 | 226,745 | 4 | -0.547 | -0.653 | 2.188 |
| chr3 | 42,307,519 | 42,307,866 | 0.024 | 0.097 | 151,540 | 303,761 | 303,764 | 4 | 0.544 | 0.672 | 2.175 |
| chr1 | 159,825,552 | 159,825,761 | 0.028 | 0.112 | 16,043 | 32,151 | 32,154 | 4 | 0.540 | 0.740 | 2.160 |
| chr8 | 54,569,668 | 54,570,293 | 0.031 | 0.124 | 217,968 | 440,066 | 440,069 | 4 | 0.536 | 0.569 | 2.143 |
| chr5 | 493,262 | 493,746 | 0.036 | 0.182 | 172,643 | 343,608 | 343,612 | 5 | 0.424 | 0.485 | 2.121 |
| chr6 | 29,795,501 | 29,795,595 | 0.034 | 0.136 | 188,094 | 374,395 | 374,398 | 4 | 0.525 | 0.586 | 2.101 |
| chr17 | 80,289,500 | 80,289,701 | -0.027 | 0.107 | 104,929 | 210,429 | 210,432 | 4 | -0.520 | -0.603 | 2.081 |
| chr10 | 70,321,770 | 70,321,959 | 0.031 | 0.124 | 28,175 | 54,833 | 54,836 | 4 | 0.519 | 0.636 | 2.075 |
| chr12 | 117,797,056 | 117,797,635 | 0.040 | 0.200 | 57,778 | 115,485 | 115,489 | 5 | 0.413 | 0.545 | 2.064 |
| chr19 | 11,353,961 | 11,354,240 | 0.028 | 0.113 | 111,615 | 224,525 | 224,528 | 4 | 0.514 | 0.652 | 2.055 |
| chr2 | 26,624,760 | 26,624,865 | 0.025 | 0.101 | 121,960 | 246,995 | 246,998 | 4 | 0.512 | 0.579 | 2.048 |
| chr16 | 56,696,748 | 56,697,229 | 0.028 | 0.111 | 88,038 | 174,840 | 174,843 | 4 | 0.511 | 0.680 | 2.045 |
| chr4 | 140,656,749 | 140,657,110 | 0.026 | 0.103 | 169,748 | 338,243 | 338,246 | 4 | 0.510 | 0.631 | 2.040 |
| chr19 | 1,387,394 | 1,387,894 | -0.028 | 0.110 | 108,823 | 218,581 | 218,584 | 4 | -0.509 | -0.687 | 2.036 |
| chr14 | 102,554,826 | 102,554,977 | 0.033 | 0.132 | 72,843 | 145,414 | 145,417 | 4 | 0.509 | 0.647 | 2.035 |
| chr6 | 10,883,895 | 10,884,314 | 0.033 | 0.130 | 186,038 | 369,196 | 369,199 | 4 | 0.508 | 0.553 | 2.034 |
| chr17 | 56,565,286 | 56,565,644 | 0.027 | 0.109 | 100,960 | 201,742 | 201,745 | 4 | 0.506 | 0.511 | 2.025 |
| chr20 | 44,746,392 | 44,747,006 | -0.028 | 0.285 | 140,615 | 282,425 | 282,434 | 10 | -0.202 | -0.249 | 2.018 |
| chr6 | 146,755,301 | 146,755,900 | 0.027 | 0.110 | 196,115 | 396,635 | 396,638 | 4 | 0.500 | 0.767 | 1.999 |
| chr1 | 11,708,792 | 11,709,271 | 0.033 | 0.131 | 3,510 | 7,279 | 7,282 | 4 | 0.496 | 0.660 | 1.983 |

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|--------------|-------------|-------------|--------|-------|---------|---------|---------|---|--------|--------|-------|
| chr11 | 82,443,149 | 82,443,614 | 0.028 | 0.113 | 44,506 | 89,617 | 89,620 | 4 | 0.496 | 0.675 | 1.982 |
| chr1 | 17,215,834 | 17,216,201 | 0.037 | 0.147 | 4,452 | 8,979 | 8,982 | 4 | 0.494 | 0.646 | 1.975 |
| chr2 | 233,251,770 | 233,252,170 | 0.041 | 0.166 | 135,805 | 272,015 | 272,018 | 4 | 0.483 | 0.516 | 1.933 |
| chr1 | 23,884,703 | 23,885,086 | 0.035 | 0.139 | 5,630 | 11,094 | 11,097 | 4 | 0.482 | 0.511 | 1.930 |
| chr8 | 143,763,326 | 143,763,565 | -0.028 | 0.110 | 223,182 | 449,333 | 449,336 | 4 | -0.482 | -0.511 | 1.927 |
| chr8 | 145,008,957 | 145,009,406 | -0.030 | 0.118 | 223,731 | 450,584 | 450,587 | 4 | -0.481 | -0.548 | 1.925 |
| chr1 | 7,842,159 | 7,842,406 | -0.033 | 0.132 | 2,675 | 5,804 | 5,807 | 4 | -0.480 | -0.577 | 1.919 |
| chr20 | 61,732,467 | 61,732,608 | 0.027 | 0.106 | 141,955 | 285,308 | 285,311 | 4 | 0.476 | 0.561 | 1.904 |
| chr22 | 51,016,501 | 51,016,644 | 0.025 | 0.100 | 148,747 | 298,687 | 298,690 | 4 | 0.471 | 0.574 | 1.885 |
| chr19 | 52,996,083 | 52,996,617 | 0.029 | 0.117 | 118,026 | 238,929 | 238,932 | 4 | 0.469 | 0.534 | 1.877 |
| chr1 | 227,748,424 | 227,748,719 | 0.030 | 0.120 | 21,322 | 41,802 | 41,805 | 4 | 0.466 | 0.588 | 1.863 |
| chr3 | 50,487,955 | 50,488,230 | 0.025 | 0.098 | 152,798 | 306,618 | 306,621 | 4 | 0.464 | 0.620 | 1.856 |
| chr13 | 112,712,424 | 112,712,795 | 0.025 | 0.101 | 65,563 | 130,953 | 130,956 | 4 | 0.458 | 0.570 | 1.832 |
| chr1 | 228,346,014 | 228,346,347 | 0.028 | 0.112 | 21,442 | 42,079 | 42,082 | 4 | 0.458 | 0.540 | 1.830 |
| chr19 | 19,639,553 | 19,639,596 | 0.033 | 0.131 | 113,496 | 228,856 | 228,859 | 4 | 0.455 | 0.615 | 1.819 |
| chr22 | 46,449,498 | 46,449,821 | -0.031 | 0.124 | 147,948 | 297,262 | 297,265 | 4 | -0.453 | -0.486 | 1.811 |
| chr2 | 114,033,360 | 114,033,830 | 0.028 | 0.112 | 128,333 | 258,706 | 258,709 | 4 | 0.448 | 0.454 | 1.792 |
| chr20 | 5,485,144 | 5,485,294 | -0.030 | 0.181 | 138,494 | 277,554 | 277,559 | 6 | -0.298 | -0.320 | 1.787 |
| chr8 | 56,791,576 | 56,791,798 | -0.033 | 0.132 | 218,114 | 440,360 | 440,363 | 4 | -0.446 | -0.476 | 1.785 |
| chr6 | 31,148,404 | 31,148,483 | -0.025 | 0.124 | 188,606 | 377,487 | 377,491 | 5 | -0.349 | -0.373 | 1.747 |
| chr13 | 112,187,145 | 112,187,396 | 0.030 | 0.121 | 65,453 | 130,675 | 130,678 | 4 | 0.419 | 0.484 | 1.676 |
| chr17 | 73,584,029 | 73,584,111 | 0.028 | 0.141 | 102,695 | 205,201 | 205,205 | 5 | 0.335 | 0.522 | 1.673 |
| chr20 | 44,803,246 | 44,803,686 | 0.032 | 0.128 | 140,625 | 282,446 | 282,449 | 4 | 0.416 | 0.476 | 1.663 |

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|--------------|-------------|-------------|--------|-------|---------|---------|---------|---|--------|--------|-------|
| chr15 | 81,410,745 | 81,411,066 | 0.031 | 0.126 | 79,970 | 158,935 | 158,938 | 4 | 0.416 | 0.507 | 1.662 |
| chr7 | 27,138,712 | 27,138,974 | 0.026 | 0.105 | 202,131 | 409,513 | 409,516 | 4 | 0.415 | 0.493 | 1.661 |
| chr6 | 32,055,135 | 32,055,316 | 0.027 | 0.107 | 188,977 | 380,461 | 380,464 | 4 | 0.411 | 0.627 | 1.643 |
| chr12 | 6,486,598 | 6,486,709 | -0.024 | 0.098 | 49,523 | 99,109 | 99,112 | 4 | -0.411 | -0.567 | 1.643 |
| chr12 | 53,358,946 | 53,359,506 | 0.030 | 0.118 | 53,044 | 105,984 | 105,987 | 4 | 0.410 | 0.466 | 1.640 |
| chr12 | 132,939,657 | 132,939,992 | 0.028 | 0.114 | 60,038 | 120,334 | 120,337 | 4 | 0.409 | 0.455 | 1.638 |
| chr2 | 66,659,348 | 66,659,590 | 0.025 | 0.098 | 124,840 | 252,149 | 252,152 | 4 | 0.404 | 0.514 | 1.615 |
| chr16 | 54,321,848 | 54,322,494 | 0.027 | 0.108 | 87,801 | 174,341 | 174,344 | 4 | 0.401 | 0.574 | 1.602 |
| chr17 | 43,716,423 | 43,716,617 | -0.024 | 0.098 | 99,443 | 198,608 | 198,611 | 4 | -0.393 | -0.451 | 1.570 |
| chr5 | 496,069 | 496,476 | 0.035 | 0.141 | 172,645 | 343,618 | 343,621 | 4 | 0.392 | 0.628 | 1.568 |
| chr9 | 96,715,687 | 96,716,209 | 0.025 | 0.101 | 226,639 | 455,043 | 455,046 | 4 | 0.386 | 0.424 | 1.543 |
| chr4 | 80,885,735 | 80,886,264 | 0.024 | 0.097 | 167,324 | 333,738 | 333,741 | 4 | 0.378 | 0.548 | 1.514 |
| chr6 | 37,616,410 | 37,616,803 | 0.027 | 0.107 | 190,315 | 385,855 | 385,858 | 4 | 0.378 | 0.556 | 1.512 |
| chr16 | 4,103,167 | 4,103,533 | -0.026 | 0.103 | 84,029 | 167,363 | 167,366 | 4 | -0.372 | -0.470 | 1.486 |
| chr16 | 55,794,456 | 55,794,910 | 0.033 | 0.164 | 87,927 | 174,589 | 174,593 | 5 | 0.272 | 0.363 | 1.361 |
| chr9 | 34,370,781 | 34,370,894 | 0.025 | 0.099 | 224,994 | 452,784 | 452,787 | 4 | 0.336 | 0.491 | 1.345 |
| chr22 | 26,875,499 | 26,875,652 | -0.025 | 0.102 | 145,844 | 292,777 | 292,780 | 4 | -0.336 | -0.374 | 1.342 |
| chr4 | 940,614 | 941,054 | 0.029 | 0.115 | 162,328 | 323,947 | 323,950 | 4 | 0.333 | 0.414 | 1.330 |
| chr7 | 4,901,337 | 4,901,628 | 0.026 | 0.105 | 200,602 | 406,489 | 406,492 | 4 | 0.330 | 0.366 | 1.318 |
| chr17 | 81,045,495 | 81,045,863 | 0.025 | 0.101 | 105,350 | 211,445 | 211,448 | 4 | 0.320 | 0.590 | 1.281 |
| chr6 | 292,329 | 292,823 | -0.031 | 0.157 | 184,828 | 366,824 | 366,828 | 5 | -0.250 | -0.260 | 1.252 |
| chr7 | 4,832,112 | 4,832,359 | 0.030 | 0.119 | 200,565 | 406,401 | 406,404 | 4 | 0.309 | 0.356 | 1.236 |
| chr5 | 139,227,979 | 139,228,242 | 0.038 | 0.192 | 180,529 | 358,423 | 358,427 | 5 | 0.247 | 0.311 | 1.236 |

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|--------------|-------------|-------------|--------|-------|---------|---------|---------|---|--------|--------|-------|
| chr11 | 2,406,712 | 2,407,267 | -0.027 | 0.109 | 36,227 | 72,232 | 72,235 | 4 | -0.292 | -0.551 | 1.168 |
| chr11 | 6,592,066 | 6,592,745 | 0.024 | 0.096 | 36,931 | 73,912 | 73,915 | 4 | 0.289 | 0.321 | 1.157 |
| chr5 | 664,363 | 664,666 | -0.035 | 0.139 | 172,747 | 343,836 | 343,839 | 4 | -0.287 | -0.317 | 1.147 |
| chr3 | 194,705,841 | 194,706,168 | -0.030 | 0.121 | 161,493 | 322,192 | 322,195 | 4 | -0.274 | -0.334 | 1.096 |
| chr21 | 46,975,805 | 46,976,340 | 0.027 | 0.110 | 144,394 | 290,012 | 290,015 | 4 | 0.264 | 0.404 | 1.055 |
| chr6 | 31,237,029 | 31,237,405 | 0.045 | 0.179 | 188,622 | 377,561 | 377,564 | 4 | 0.232 | 0.238 | 0.928 |
| chr19 | 55,477,653 | 55,477,810 | -0.030 | 0.120 | 118,579 | 240,113 | 240,116 | 4 | -0.164 | -0.198 | 0.655 |
| chr2 | 128,453,108 | 128,453,484 | 0.034 | 0.171 | 129,225 | 260,338 | 260,342 | 5 | 0.114 | 0.157 | 0.571 |
| chr4 | 6,728,936 | 6,729,199 | 0.035 | 0.141 | 163,790 | 327,281 | 327,284 | 4 | 0.128 | 0.205 | 0.513 |
| chr12 | 131,622,284 | 131,622,739 | -0.028 | 0.111 | 59,596 | 119,302 | 119,305 | 4 | -0.085 | -0.163 | 0.340 |

The table shows the results of the *dmrFind* algorithm. DMR positions are shown in the chr/start/end co-ordinates, with probe indices and numbers represented by indexStart, indexEnd and nprobes, and area_raw the significance calculation following permutation analysis, allowing ranking of these DMRs by significance, as shown.

Weighted Gene Co-expression Network Analysis (WGCNA)

We performed weighted gene co-expression network analysis (WGCNA) (Langfelder and Horvath 2008) to assess networks of co-methylated CpGs associated with ASD status. We used the CpG values output from the surrogate variable analysis (SVA) algorithm called in bump-hunting, since SVA corrected for all known technical artifacts. We used the WGCNA package in R and built an unsigned co-methylation network. Correlation matrices were raised to the power of 5, as calculated by the scale-free topology criterion on data subsets, and thresholds were set of minimum module connectivity (kME) of greater than 0.7, and minimum height for module merging of 0.1 (Voineagu et al. 2011). We ran the algorithm with block sizes of 40,000 CpGs.

We assessed module relevance to case/control status with a t-test (two tailed, unequal variance) of module eigengene values with case/control and gender categories. For relationship with the continuous variables of age and percent YRI and CEU ancestry, we used Pearson correlation coefficients and their Student asymptotic p-values.

For analysis of methylation changes associated with ASD, we selected the 2 modules (“light green”, and “dark olive green2”) that showed significant correlation only with ASD status and not with any other covariate, to avoid confounding effects.

A full list of the modules obtained and their Bonferroni-corrected p-values is provided in **Supplemental Table S7**.

The genes associated with “light green” and “dark olive green 2” two ASD-associated modules are listed in **Supplemental Tables S8 and S9**.

Gene ontology enrichment was performed using the same Cytoscape method described previously. For the light green module, gene ontology showed significant enrichment for negative regulation of smooth muscle cell migration ($p = 1.34 \times 10^{-2}$), regulation of cell proliferation ($p = 3.25 \times 10^{-2}$), negative regulation of cell migration ($p = 3.27 \times 10^{-2}$), negative regulation of locomotion ($p = 3.27 \times 10^{-2}$), negative regulation of cellular component movement ($p = 3.27 \times 10^{-2}$), and negative regulation of metabolic process ($p = 4.64 \times 10^{-2}$). All p-values shown are after FDR correction.

It has been recently demonstrated that gene ontology analysis can be affected by unequal probe distribution in microarrays, with bias towards genes represented by greater numbers of probes (Geeleher et al. 2013). To address this concern, we calculated the number of probes corresponding to every gene by using annotations assigned by Illumina in the 450K array manifest. We used the number of probes per gene to generate a weight for every gene and recalculated enriched gene ontologies, using the R package *GoSeq* (Young et al. 2010). After accounting for unequal probe distribution, we obtained candidate ontology categories, but these did not remain significant after correcting the associated p-values for multiple testing.

We also interrogated some of the modules most significantly associated with age and ancestry, the “dark turquoise” and “red” modules, respectively. Using the *Lists2Networks* software (Lachmann and Ma'ayan 2010), we found that the most significant gene ontology category enriched with genes from the age module was brain development ($p=0.008$). We then asked whether the ancestry-associated modules were enriched for CpGs with annotated SNPs on either the C or G. Out of 247 CpGs, 154 contained a reported variant (based on 1000 Genomes data), with 11 of those possessing variants on both the C and the G. This confirms the

importance of both polymorphism on the CpG, as well as the presence of ancestry-associated methylation changes resulting from effects other than direct polymorphism.

Protein-Protein Interaction (PPI) Analysis

To assess the functional impact of our WGCNA ASD-associated co-methylated gene modules, we interrogated the genes' relevance in protein-protein interaction (PPI) networks. We combined the genes in the light green and dark olive green 2 modules with a list of previously curated known ASD risk genes, with the addition of exome sequencing candidates (*KATNAL2* and *CHD8*) (Neale et al. 2012). We used GeneMania (Warde-Farley et al. 2010) to build a PPI network of this combined list, using only data from physical protein interaction databases. We visualized this network in Cytoscape.

To test the significance of these PPI connections, we performed Degree Aware Disease Gene Prioritization (DADA) (Erten et al. 2011). We used the ASD seed list mentioned previously, a combined candidate list of the genes related to the ASD-associated WGCNA CpGs, and the physical interaction database from the Human Protein Reference Database (HPRD) available through GeneMania. We evaluated if our genes were significantly enriched in ranking using the Mann-Whitney test.

We also assessed whether the candidate genes related to ASD-associated CpG modules were functionally connected to known risk genes for Intellectual Disability (ID). We used a previously curated list of ID-implicated genes (Neale et al. 2012) and combined that with our 2 module genes to build a PPI network. Using the same parameters as above, we generated a network showing many functional connections, displayed in **Supplemental Figure S8**.



Supplemental Table S7: Bonferroni-corrected p-values of association between modules and known covariates

| Module | ASD | Gender | Age | YRI % | CEU % |
|-------------------|----------|--------|----------|----------|-------------|
| MElavenderblush2 | 1 | 1 | 1 | 1 | 1 |
| Meplum3 | 1 | 1 | 1 | 1 | 1 |
| Mecoral2 | 1 | 1 | 1 | 1 | 1 |
| Methistle3 | 1 | 1 | 1 | 1 | 1 |
| Memediumorchid | 1 | 1 | 1 | 1 | 1 |
| Medeepink | 1 | 1 | 1 | 1 | 1 |
| Melightslateblue | 1 | 1 | 1 | 1 | 1 |
| Mepaleturquoise | 1 | 1 | 1 | 1 | 1 |
| Mepalevioletred3 | 1 | 1 | 1.38E-02 | 1 | 1 |
| Mechocolate4 | 1 | 1 | 3.18E-04 | 1 | 1 |
| Memediumpurple1 | 1 | 1 | 1 | 1 | 1 |
| Menavajowhite1 | 1 | 1 | 9.65E-02 | 1 | 1 |
| Medarkseagreen3 | 1 | 1 | 4.47E-02 | 1 | 1 |
| Mepink4 | 1 | 1 | 7.86E-02 | 1 | 1 |
| Mehoneydew | 1 | 1 | 5.09E-04 | 1 | 1 |
| Mesaddlebrown | 0.65 | 1 | 3.67E-03 | 1 | 1 |
| Mepalevioletred2 | 1 | 1 | 3.04E-03 | 1 | 1 |
| Methistle1 | 2.43E-02 | 1 | 7.58E-05 | 1 | 1 |
| Memagenta4 | 0.16 | 1 | 3.09E-17 | 1 | 1 |
| Meyellow | 8.50E-04 | 1 | 8.40E-16 | 2.53E-02 | 1.98E-02 |
| Mefirebrick3 | 8.23E-02 | 1 | 2.31E-08 | 1 | 0.88 |
| Mepalevioletred1 | 2.85E-02 | 1 | 1.73E-10 | 2.06E-02 | 1.38E-02 |
| Meplum2 | 1 | 1 | 4.00E-09 | 1 | 1 |
| Melightsteelblue1 | 1 | 1 | 3.61E-15 | 0.59 | 0.74 |
| Menavajowhite2 | 0.21 | 1 | 5.86E-11 | 0.33 | 0.51 |
| Mecoral3 | 1 | 1 | 4.13E-15 | 1 | 1 |
| Mebrown | 9.93E-03 | 1 | 9.61E-28 | 1.27E-02 | 1.40E-02 |
| Meskyblue1 | 1 | 1 | 3.32E-23 | 1 | 1 |
| Mecoral4 | 2.31E-02 | 1 | 2.30E-03 | 1 | 1 |
| Mecyan | 1 | 1 | 1 | 1 | 1 |
| Metan4 | 5.92E-02 | 1 | 1.55E-04 | 1 | 1 |
| Mehoneydew1 | 1 | 1 | 3.28E-02 | 1 | 1 |
| Melightcoral | 1 | 1 | 1.12E-04 | 0.23 | 1 |
| Meorange | 1 | 1 | 1.64E-08 | 9.54E-02 | 0.280779828 |
| Meantiquewhite2 | 1 | 1 | 1 | 1 | 1 |
| Megreen | 1 | 1 | 9.28E-03 | 1 | 1 |
| Meyellow4 | 1 | 1 | 4.17E-06 | 1 | 1 |
| Meblue | 1 | 1 | 3.95E-06 | 1 | 1 |
| Meturquoise | 1 | 1 | 8.25E-06 | 1 | 1 |
| Medarkseagreen4 | 1 | 1 | 1 | 1 | 1 |
| Medarkgreen | 1 | 1 | 1 | 1 | 1 |

| | | | | | |
|--------------------------|-----------------|----------|-------------|-------------|-------------|
| Mefloralwhite | 1 | 0.92 | 1 | 6.57E-03 | 2.44E-04 |
| Meorangered4 | 1 | 1 | 1 | 1 | 1 |
| Meblue4 | 1 | 1 | 1 | 1 | 1 |
| Megreen4 | 1 | 1 | 1 | 1 | 1 |
| Melightsteelblue | 1 | 1 | 1 | 1 | 1 |
| Meskyblue3 | 1 | 1 | 1 | 1 | 1 |
| Mesalmon2 | 1 | 1 | 1 | 1 | 1 |
| Meskyblue2 | 1 | 1 | 1 | 1 | 1 |
| Mesienna3 | 1 | 1 | 1 | 1 | 1 |
| Meorangered3 | 1 | 1 | 1 | 1 | 1 |
| Meplum1 | 1 | 1 | 1 | 1 | 1 |
| Meyellowgreen | 1 | 1 | 1 | 1 | 1 |
| Mesienna4 | 1 | 1 | 1 | 1 | 1 |
| Mesteelblue | 1 | 1 | 1 | 1 | 1 |
| Meindianred4 | 1 | 1 | 1 | 1 | 1 |
| Melavenderblush1 | 1 | 1 | 1 | 1 | 1 |
| Medarkslateblue | 1 | 1 | 1 | 1 | 1 |
| Meskyblue4 | 1 | 1 | 1 | 1 | 1 |
| Mesalmon1 | 1 | 1 | 1 | 1 | 1 |
| Mewhite | 1 | 1 | 1 | 1 | 1 |
| Memidnightblue | 1 | 1 | 1 | 1 | 1 |
| Methistle2 | 1 | 1 | 1 | 1 | 1 |
| Medarkviolet | 1 | 1 | 1 | 1 | 1 |
| Melightgreen | 5.26E-02 | 1 | 1 | 1 | 1 |
| Meskyblue | 1 | 1 | 1 | 1 | 1 |
| Meyellow3 | 1 | 1 | 1 | 1 | 1 |
| Medarkseagreen2 | 1 | 1 | 1 | 1 | 1 |
| Meorangered1 | 1 | 1 | 1 | 1 | 1 |
| Meivory | 1 | 1 | 1 | 1 | 1 |
| Medarkolivegreen | 1 | 1 | 1 | 1 | 1 |
| Memagenta3 | 1 | 1 | 1 | 1 | 1 |
| Melightcyan | 1 | 1 | 1 | 4.30E-02 | 0.59 |
| Melighpink4 | 1 | 1 | 1 | 1 | 1 |
| Meroyalblue | 1 | 1 | 1 | 1 | 0.10 |
| Mecoral | 1 | 1 | 1 | 1 | 1 |
| Medarkgrey | 1 | 1 | 1 | 1 | 1 |
| Memagenta | 1 | 1 | 1 | 0.19 | 0.64 |
| Melavenderblush3 | 1 | 1 | 1 | 1 | 1 |
| Mefirebrick4 | 1 | 1 | 1 | 1 | 1 |
| Methistle4 | 1 | 1 | 0.28 | 1 | 1 |
| Medarkmagenta | 1 | 1 | 1 | 0.67 | 0.24 |
| Medarkolivegreen2 | 1.01E-02 | 1 | 0.18 | 0.42 | 0.54 |
| Memediumpurple2 | 1 | 1 | 1 | 1 | 1 |
| Meviolet | 1 | 1 | 1 | 1 | 1 |

| | | | | | |
|-------------------|----------|----------|----------|----------|------------|
| Meblack | 1 | 1 | 1 | 5.83E-02 | 0.47 |
| Megreenyellow | 1 | 1 | 1 | 3.28E-03 | 3.61E-02 |
| Memaroon | 1 | 0.64 | 1 | 0.59 | 0.82 |
| Mepurple | 0.51 | 1.08E-42 | 1 | 1 | 1 |
| Meantiquewhite4 | 1 | 1 | 1 | 1 | 1 |
| Meblueviolet | 1 | 1 | 1 | 1 | 1 |
| Meplum4 | 1 | 1 | 1 | 1 | 1 |
| Memediumpurple3 | 1 | 1 | 1 | 1 | 1 |
| Menavajowhite | 1 | 1 | 1 | 1 | 1 |
| Mesalmon4 | 1 | 1 | 1 | 1 | 1 |
| Meindianred3 | 1 | 1 | 1 | 1 | 1 |
| Meblue2 | 1 | 1 | 1 | 1 | 1 |
| Mecoral1 | 1 | 1 | 1 | 1 | 1 |
| Meplum | 1 | 1 | 1 | 1 | 1 |
| Mered | 1 | 1 | 1 | 5.97E-03 | 3.75E-02 |
| Mesalmon | 1 | 1 | 1 | 6.59E-02 | 0.39877902 |
| Melightblue4 | 1 | 1 | 1 | 1 | 1 |
| Mebrown2 | 0.91 | 1 | 1 | 1 | 1 |
| Medarkturquoise | 4.07E-02 | 1 | 8.32E-29 | 1 | 1 |
| Melightlyellow | 3.68E-02 | 1 | 2.75E-20 | 1 | 1 |
| Medarkorange2 | 1 | 1 | 6.18E-04 | 1 | 1 |
| Metan | 1 | 1 | 2.51E-09 | 1 | 1 |
| Melightcyan1 | 1 | 1 | 6.23E-02 | 1 | 1 |
| Mebisque4 | 1 | 1 | 1 | 1 | 1 |
| Memediumpurple4 | 1 | 1 | 1 | 1 | 1 |
| Meantiquewhite1 | 1 | 1 | 1.42E-03 | 1 | 1 |
| Medarkolivegreen4 | 0.38 | 1 | 1.13E-05 | 0.99 | 0.47 |
| Medarkorange | 2.48E-02 | 1 | 1.26E-07 | 1 | 1 |
| Melighpink3 | 1 | 1 | 1 | 1 | 1 |
| Medarkred | 1 | 1 | 1 | 1 | 1 |
| Megrey60 | 1 | 1 | 1 | 0.19 | 1 |
| Mebrown4 | 1 | 1 | 1 | 1 | 1 |
| Mepink | 1 | 1 | 1 | 2.08E-02 | 0.15 |
| Melighpink2 | 1 | 1 | 1 | 1 | 1 |
| Methistle | 1 | 1 | 1.75E-02 | 1 | 1 |
| Megrey | 6.01E-05 | 1 | 2.81E-30 | 4.72E-03 | 3.28E-03 |

Supplemental Table S8: Genes in *light green* module associated with ASD

| Gene | Chromosome | Position | Probe | Module Membership (MM) | MM P-value |
|----------|------------|-----------|----------------|------------------------|------------|
| HES4 | 1 | 936610 | cg15825501 | 0.80 | 6.35E-22 |
| ALDH4A1 | 1 | 19229528 | cg01972394 | 0.76 | 9.95E-19 |
| GRIK3 | 1 | 37498270 | cg04364463 | 0.72 | 8.16E-16 |
| DPH2 | 1 | 44435457 | cg06790019 | 0.70 | 1.47E-14 |
| EIF2B3 | 1 | 45452166 | cg05654404 | 0.73 | 9.48E-17 |
| RAD54L | 1 | 46712932 | cg16163324 | 0.79 | 1.18E-20 |
| C1orf183 | 1 | 112281877 | cg21304211 | 0.88 | 2.41E-31 |
| ST7L | 1 | 113162073 | cg17861791 | 0.77 | 2.17E-19 |
| CAPZA1 | 1 | 113162073 | cg17861791 | 0.77 | 2.17E-19 |
| DENND2C | 1 | 115212659 | cg13501090 | 0.85 | 8.00E-27 |
| LGR6 | 1 | 202162209 | cg04351903 | 0.85 | 5.85E-27 |
| PTPN14 | 1 | 214725274 | cg17024643 | 0.78 | 8.04E-20 |
| MORN2 | 2 | 39103277 | cg07479988 | 0.79 | 1.38E-20 |
| DHX57 | 2 | 39103277 | cg07479988 | 0.79 | 1.38E-20 |
| MGAT5 | 2 | 134949571 | cg16240751 | 0.76 | 1.67E-18 |
| RIF1 | 2 | 152266336 | cg18320648 | 0.80 | 9.07E-22 |
| RPE | 2 | 210867059 | cg10181911 | 0.72 | 3.05E-16 |
| IGFBP5 | 2 | 217559020 | cg03222971 | 0.74 | 2.99E-17 |
| SCAP | 3 | 47516975 | cg01374398 | 0.77 | 4.27E-19 |
| TNNC1 | 3 | 52489346 | cg01910272 | 0.71 | 3.55E-15 |
| NISCH | 3 | 52489346 | cg01910272 | 0.71 | 3.55E-15 |
| MAGI1 | 3 | 66024691 | cg15764058 | 0.72 | 6.66E-16 |
| NAALADL2 | 3 | 174095241 | cg17014718 | -0.71 | 1.86E-15 |
| HES1 | 3 | 193852754 | cg26348180 | 0.76 | 2.91E-18 |
| MAN2B2 | 4 | 6577027 | cg09477292 | 0.76 | 2.38E-18 |
| HS3ST1 | 4 | 11810667 | ch.4.11419765R | 0.76 | 2.21E-18 |
| CD38 | 4 | 15780522 | cg27473538 | 0.73 | 1.31E-16 |
| RBPJ | 4 | 26323246 | cg06812693 | 0.82 | 4.72E-23 |
| FRAS1 | 4 | 78977690 | cg12573119 | 0.76 | 1.78E-18 |
| FRAS1 | 4 | 78978133 | cg01402409 | 0.74 | 3.84E-17 |
| COQ2 | 4 | 84205961 | cg20161984 | 0.77 | 2.09E-19 |
| LARP7 | 4 | 113626831 | cg03148140 | 0.71 | 1.34E-15 |
| SMAD1 | 4 | 146296778 | cg06295548 | -0.72 | 6.74E-16 |
| HMGB2 | 4 | 174254825 | cg14705778 | 0.73 | 1.90E-16 |
| RGS7BP | 5 | 63802184 | cg08709073 | 0.78 | 4.86E-20 |
| FAM151B | 5 | 79783889 | cg06506598 | 0.72 | 9.98E-16 |
| PRDM6 | 5 | 122435202 | cg01196322 | 0.75 | 5.20E-18 |
| NEUROG1 | 5 | 134871686 | cg17772342 | 0.72 | 3.06E-16 |
| SPARC | 5 | 151031796 | cg21530174 | 0.72 | 3.59E-16 |
| TSPAN17 | 5 | 176131088 | cg13041389 | -0.79 | 7.70E-21 |
| ZFP62 | 5 | 180287685 | cg24877792 | 0.80 | 2.45E-21 |
| LY86 | 6 | 6648823 | cg16101278 | -0.70 | 6.61E-15 |

| | | | | | |
|-------------------|----|-----------|------------|-------|----------|
| <i>BAT4</i> | 6 | 31634141 | cg27137280 | 0.70 | 5.78E-15 |
| <i>CSNK2B</i> | 6 | 31634141 | cg27137280 | 0.70 | 5.78E-15 |
| <i>PPIL1</i> | 6 | 36842651 | cg04860157 | 0.81 | 7.24E-23 |
| <i>CRIP3</i> | 6 | 43276478 | cg18857062 | 0.74 | 2.91E-17 |
| <i>LCA5</i> | 6 | 80246572 | cg19879479 | 0.84 | 5.79E-26 |
| <i>NDUFAF4</i> | 6 | 97345972 | cg04571327 | 0.70 | 9.24E-15 |
| <i>C6orf174</i> | 6 | 127837548 | cg21986718 | 0.75 | 6.74E-18 |
| <i>TNRC18</i> | 7 | 5463409 | cg23209537 | 0.71 | 1.93E-15 |
| <i>IGFBP3</i> | 7 | 45961943 | cg15208757 | 0.76 | 3.38E-18 |
| <i>TYW1B</i> | 7 | 72298667 | cg21423973 | 0.72 | 4.62E-16 |
| <i>SBDSP</i> | 7 | 72298667 | cg21423973 | 0.72 | 4.62E-16 |
| <i>GATSL1</i> | 7 | 74379144 | cg18210722 | -0.69 | 1.63E-14 |
| <i>LHFPL3</i> | 7 | 103969483 | cg17373058 | 0.71 | 2.38E-15 |
| <i>LHFPL3</i> | 7 | 103970195 | cg11826638 | 0.70 | 5.50E-15 |
| <i>TSPAN12</i> | 7 | 120497479 | cg26340461 | 0.72 | 3.42E-16 |
| <i>RHEB</i> | 7 | 151215566 | cg20495206 | 0.80 | 7.33E-22 |
| <i>SORBS3</i> | 8 | 22423994 | cg21291431 | 0.78 | 1.07E-19 |
| <i>TNFRSF10B</i> | 8 | 22926800 | cg26918957 | 0.71 | 2.51E-15 |
| <i>DPYSL2</i> | 8 | 26434689 | cg14967899 | 0.72 | 5.60E-16 |
| <i>RRM2B</i> | 8 | 103251909 | cg12374732 | 0.70 | 1.30E-14 |
| <i>MYC</i> | 8 | 128748155 | cg25080152 | 0.73 | 1.62E-16 |
| <i>PCSK5</i> | 9 | 78506874 | cg13512204 | 0.79 | 3.30E-21 |
| <i>ZEB1</i> | 10 | 31608136 | cg25231972 | 0.75 | 1.02E-17 |
| <i>FZD8</i> | 10 | 35930499 | cg00645593 | 0.72 | 3.60E-16 |
| <i>ZNF503-AS1</i> | 10 | 77054788 | cg15394763 | 0.70 | 5.06E-15 |
| <i>FGF8</i> | 10 | 103535362 | cg11706469 | 0.78 | 1.15E-19 |
| <i>ZNF215</i> | 11 | 6948101 | cg10765857 | 0.80 | 9.54E-22 |
| <i>GTF2H1</i> | 11 | 18343657 | cg11347316 | 0.74 | 4.99E-17 |
| <i>HPS5</i> | 11 | 18343657 | cg11347316 | 0.74 | 4.99E-17 |
| <i>PAX6</i> | 11 | 31831591 | cg16822387 | 0.72 | 7.24E-16 |
| <i>KBTBD4</i> | 11 | 47600851 | cg07996345 | 0.73 | 1.87E-16 |
| <i>NDUFS3</i> | 11 | 47600851 | cg07996345 | 0.73 | 1.87E-16 |
| <i>FADS1</i> | 11 | 61584442 | cg25837350 | 0.76 | 2.55E-18 |
| <i>VEGFB</i> | 11 | 64002754 | cg18872604 | 0.70 | 1.51E-14 |
| <i>BAD</i> | 11 | 64052221 | cg25163015 | 0.79 | 6.58E-21 |
| <i>GPR137</i> | 11 | 64052221 | cg25163015 | 0.79 | 6.58E-21 |
| <i>KRTAP5-11</i> | 11 | 71340352 | cg22870994 | -0.81 | 2.92E-22 |
| <i>GAB2</i> | 11 | 78129288 | cg05492810 | 0.77 | 4.88E-19 |
| <i>TMEM126B</i> | 11 | 85339628 | cg12830327 | 0.78 | 2.11E-20 |
| <i>DLG2</i> | 11 | 85339628 | cg12830327 | 0.78 | 2.11E-20 |
| <i>MPZL3</i> | 11 | 118123074 | cg27161463 | 0.71 | 2.29E-15 |
| <i>CLEC4C</i> | 12 | 7904267 | cg18348303 | -0.73 | 7.50E-17 |
| <i>DDX11</i> | 12 | 31226536 | cg16864700 | 0.75 | 1.16E-17 |
| <i>NELL2</i> | 12 | 45270304 | cg21846305 | 0.74 | 2.41E-17 |

| | | | | | |
|-----------|----|-----------|------------|-------|----------|
| TRHDE | 12 | 72667326 | cg09972192 | 0.71 | 2.35E-15 |
| LOC283392 | 12 | 72667326 | cg09972192 | 0.71 | 2.35E-15 |
| N4BP2L1 | 13 | 33002431 | cg06513149 | 0.78 | 2.70E-20 |
| ABCC4 | 13 | 95953574 | cg15396799 | 0.75 | 1.21E-17 |
| ARHGEF7 | 13 | 111768023 | cg03925425 | 0.77 | 5.42E-19 |
| TUBGCP3 | 13 | 113263221 | cg24531141 | -0.76 | 2.00E-18 |
| EFS | 14 | 23834995 | cg02213260 | 0.75 | 4.72E-18 |
| PSME1 | 14 | 24604912 | cg24054649 | 0.71 | 2.82E-15 |
| SNX6 | 14 | 35099518 | cg13093793 | 0.74 | 6.03E-17 |
| SRP54 | 14 | 35451984 | cg04980793 | 0.70 | 5.47E-15 |
| FOXN3 | 14 | 90084672 | cg26386436 | 0.74 | 4.68E-17 |
| SLC25A29 | 14 | 100751514 | cg10963192 | 0.75 | 4.31E-18 |
| PACS2 | 14 | 105827276 | cg15936935 | -0.85 | 5.52E-27 |
| RTF1 | 15 | 41708917 | cg15581235 | 0.78 | 2.32E-20 |
| CORO2B | 15 | 68870836 | cg12043722 | 0.74 | 1.96E-17 |
| CSPG4 | 15 | 75986363 | cg14576802 | 0.71 | 2.67E-15 |
| ZNF200 | 16 | 3285262 | cg03530756 | 0.74 | 5.77E-17 |
| KLHDC4 | 16 | 87811505 | cg09562174 | 0.76 | 2.23E-18 |
| RNMTL1 | 17 | 685915 | cg27220681 | 0.71 | 1.85E-15 |
| GLOD4 | 17 | 685915 | cg27220681 | 0.71 | 1.85E-15 |
| NF1 | 17 | 29421732 | cg02726883 | 0.75 | 4.54E-18 |
| SLFN11 | 17 | 33700513 | cg05504685 | 0.72 | 5.32E-16 |
| COPZ2 | 17 | 46114574 | cg21384971 | 0.79 | 3.32E-21 |
| MIR152 | 17 | 46114574 | cg21384971 | 0.79 | 3.32E-21 |
| LOC146880 | 17 | 62777690 | cg15869463 | 0.76 | 1.34E-18 |
| KPNA2 | 17 | 66031814 | cg13777502 | 0.72 | 8.83E-16 |
| RECQL5 | 17 | 73629082 | cg04219446 | 0.74 | 3.28E-17 |
| LOC643008 | 17 | 73629082 | cg04219446 | 0.74 | 3.28E-17 |
| TIMP2 | 17 | 76921528 | cg10466987 | 0.80 | 2.29E-21 |
| CCDC165 | 18 | 8707237 | cg14095692 | 0.77 | 1.28E-19 |
| RAB27B | 18 | 52495848 | cg05095774 | 0.70 | 6.23E-15 |
| CCDC102B | 18 | 66382471 | cg15552529 | 0.78 | 3.46E-20 |
| TMX3 | 18 | 66382471 | cg15552529 | 0.78 | 3.46E-20 |
| SGTA | 19 | 2761892 | cg04171554 | 0.70 | 6.43E-15 |
| ZNF77 | 19 | 2945000 | cg04335562 | 0.80 | 5.98E-22 |
| TNPO2 | 19 | 12833533 | cg11788103 | 0.71 | 4.20E-15 |
| LYL1 | 19 | 13213716 | cg02011446 | 0.71 | 1.22E-15 |
| UQCRRF51 | 19 | 29704262 | cg02905964 | 0.74 | 6.92E-17 |
| RTN2 | 19 | 45996498 | cg19869610 | 0.74 | 6.33E-17 |
| SMOX | 20 | 4129314 | cg20604317 | 0.73 | 1.77E-16 |
| HMG1 | 21 | 40720919 | cg01338834 | 0.73 | 9.04E-17 |
| DGCR6 | 22 | 18893614 | cg07004357 | 0.71 | 1.42E-15 |
| LZTR1 | 22 | 21337040 | cg07047601 | 0.81 | 2.81E-22 |
| PNPLA5 | 22 | 44287772 | cg24258125 | 0.77 | 1.49E-19 |

Supplemental Table S9: Genes in *dark olive green* module associated with ASD

| Gene | Chromosome | Position | Probe | Module Membership (MM) | MM P-value |
|----------|------------|-----------|------------|------------------------|------------|
| PEX10 | 1 | 2337334 | cg16523185 | 0.810337844 | 1.32E-22 |
| GPBP1L1 | 1 | 46106399 | cg17827803 | 0.773502529 | 1.61E-19 |
| BCAR3 | 1 | 94245226 | cg24937735 | 0.740245636 | 3.41E-17 |
| ZC3H11A | 1 | 203763483 | cg16020436 | 0.768457607 | 3.85E-19 |
| ZC3H11A | 1 | 203763498 | cg02337583 | 0.699133215 | 9.15E-15 |
| CAMKMT | 2 | 44646510 | cg10521567 | 0.830944239 | 1.21E-24 |
| PRKCE | 2 | 46121488 | cg16884847 | 0.728533494 | 1.86E-16 |
| RBMS1 | 2 | 161593271 | cg13392885 | 0.776332535 | 9.80E-20 |
| ARPC2 | 2 | 219079038 | cg12884009 | 0.797400033 | 1.89E-21 |
| BSN | 3 | 49638532 | cg22881573 | 0.845041711 | 3.35E-26 |
| CPLX1 | 4 | 795538 | cg02133849 | 0.702505454 | 5.99E-15 |
| NFXL1 | 4 | 47854334 | cg27531236 | 0.802345605 | 7.00E-22 |
| C5orf27 | 5 | 95192949 | cg26898099 | 0.70421452 | 4.83E-15 |
| ERGIC1 | 5 | 172263112 | cg24581650 | 0.683902398 | 5.75E-14 |
| TAP2 | 6 | 32803058 | cg00386460 | 0.778753197 | 6.37E-20 |
| COL11A2 | 6 | 33151008 | cg07457375 | 0.729478157 | 1.63E-16 |
| WDR46 | 6 | 33254880 | cg23652681 | 0.801745369 | 7.91E-22 |
| WDR46 | 6 | 33254892 | cg17417645 | 0.761165479 | 1.30E-18 |
| RSPH3 | 6 | 159423743 | cg15999887 | 0.769393089 | 3.28E-19 |
| AGPAT4 | 6 | 161549519 | cg26780581 | 0.790267496 | 7.57E-21 |
| MACC1 | 7 | 20240145 | cg21710826 | 0.698674927 | 9.68E-15 |
| CUX1 | 7 | 101478535 | cg08582182 | 0.802229538 | 7.17E-22 |
| JHDM1D | 7 | 139859464 | cg26800802 | 0.713418296 | 1.47E-15 |
| KIAA0146 | 8 | 48557420 | cg04247508 | 0.792246082 | 5.18E-21 |
| KIAA0146 | 8 | 48587440 | cg18404513 | 0.753484619 | 4.48E-18 |
| PABPC1 | 8 | 101802144 | cg08995449 | 0.774123593 | 1.45E-19 |
| ARFIP2 | 11 | 6499575 | cg17403702 | 0.736923523 | 5.57E-17 |
| MICAL2 | 11 | 12222570 | cg09371112 | 0.722898685 | 4.08E-16 |
| CNIH2 | 11 | 66048759 | cg06155341 | 0.805030303 | 4.03E-22 |
| MAML2 | 11 | 95889454 | cg15521790 | 0.781796778 | 3.67E-20 |
| KRT79 | 12 | 53228661 | cg13119928 | 0.764105173 | 8.00E-19 |
| NCOR2 | 12 | 125030744 | cg07243762 | 0.722336993 | 4.41E-16 |
| ATP11A | 13 | 113348391 | cg09507215 | 0.730182655 | 1.47E-16 |
| HOMEZ | 14 | 23744304 | cg04420752 | 0.747121013 | 1.21E-17 |
| ARHGAP5 | 14 | 32597733 | cg07564690 | 0.831457161 | 1.07E-24 |
| RBM25 | 14 | 73524288 | cg04422024 | 0.757192347 | 2.48E-18 |
| c15orf50 | 15 | 70147094 | cg16497945 | 0.763106126 | 9.44E-19 |
| MLYCD | 16 | 83945978 | cg01984843 | 0.776011546 | 1.04E-19 |
| ZNF426 | 19 | 9645566 | cg21474288 | 0.749726963 | 8.08E-18 |
| PSMD8 | 19 | 38869646 | cg11607742 | 0.801710907 | 7.97E-22 |
| HM13 | 20 | 30126382 | cg04306926 | 0.728729016 | 1.81E-16 |
| PPM1F | 22 | 22290866 | cg01800253 | 0.710229687 | 2.23E-15 |

Code

Using R version 2.15.0

```
#LOADING IN RAW IDAT FILES
setwd("INSERT USER DIRECTORY")
man<-read.csv("HumanMethylation450_15017482_v.1.1.csv", skip=7, header=TRUE,
stringsAsFactors=FALSE)

require(minfi)
library(IlluminaHumanMethylation450kmanifest)
targets <- read.450k.sheet("USER DIRECTORY", pattern="pheno96.csv")
Basename<-apply(targets, 1, function(x){
  paste("USER DIRECTORY", x[13], "/", x[13], "_", x[12], sep="")
})
targets<-data.frame(targets[,1:13], Basename)
RGset <- read.450k.exp(base = "USER DIRECTORY", targets = targets)

##Extract bisulphite control values from getProbeInfo from RGset

#CALCULATE DETECTION PVALS TO REMOVE POORLY PERFORMING PROBES
sampleNames(RGset) = targets$Sample_Name
pVals = detectionP(RGset)

M.set<-preprocessRaw(RGset) #this is what the table of signal intensities is extracted from

#REMOVE SEX CHROMOSOMES BASED ON ILLUMINA MANIFEST ANNOTATION
cpgs<-getManifestInfo(M.set, type = c("locusNames"))
Y<-which(man[,12]=="Y")
X<-which(man[,12]=="X")
cpgsremove<-c(man[X,1], man[Y,1])
pos<-cpgs %in% cpgsremove
nomatch<-which(pos=="FALSE")
M.set2<-M.set[nomatch,]

#NORMALIZE AUTOSOME DATA WITH SWAN
M.setswan<-preprocessSWAN(RGset, mSet=M.set2)

#REMOVE PROBES WITH DETECTION P-VAL > 0.01
pValsauto<-pVals[nomatch,]
msetswanfilt<-M.setswan[rowSums(pValsauto)<=0.01,]

#EXTRACT M-VALUES FROM NORMALIZED, FILTERED DATASET
mvals<-getM(msetswanfilt, type="Illumina")
write.table(mvals, file="m_values.txt", sep="\t", quote=FALSE)

#CORRECT FOR BATCH EFFECT
library(sva)
pd<-pData(msetswanfilt)
```

```
batch<-pd$Slide
mod<-model.matrix(~as.factor(Gender) + age.3.2012 + as.factor(Sample_Group), data=pd)
norm_m<-ComBat(mvals, batch=batch, mod=mod, numCovs=3,
par.prior=TRUE,prior.plots=FALSE)
write.table(norm_m, file="norm_m.txt", sep="\t", quote=FALSE)

##This file contains 96 individuals. Three were then removed from subsequent DMR analysis
(one had a changed diagnosis, and two did not have genotyping data as their genotyping arrays
failed quality controls. For simplicity, the phenotype file with the 93 retained individuals is
provided for the subsequent analysis.
```

``` #BUMPHUNTING ANALYSIS ```

```
setwd("USER DIRECTORY")
mval<-read.delim("norm_m.txt", header=TRUE, stringsAsFactors=FALSE)

cpgnames<-row.names(mval)

manifest<-read.csv("HumanMethylation450_15017482_v.1.1.csv", skip=7, sep=";",
stringsAsFactors=FALSE)
temp<-match(cpgnames, manifest[,1])
manifest2<-manifest[temp,]

pheno<-read.csv("pheno93.csv", stringsAsFactors=FALSE)

names<-colnames(mval)
keep<-match(pheno[,1], names)
mval2<-mval[,keep]

chr<-manifest2[,12]
chr<-paste("chr", chr, sep="")
position<-as.numeric(manifest2[,13])
library(charm)

#this part reorders each chromosome by position and then rbinds them all together, and then
reorders the matrix of m-values by that order too)
a<-cbind(chr, position, cpgnames)
chrs<-levels(as.factor(chr))
final<-c()
for (j in 1:length(chrs)){
b<-subset(a, a[,1]==chrs[j])
ord<-order(as.numeric((b[,2])))
c<-b[ord,]
final<-rbind(final, c)
}
exclude<-which(final[,1]=="chr")
final2<-final[-exclude,]
d<-match(final2[,3], row.names(mval))
mval3<-mval2[d,]
```

```
position<-as.numeric(final2[,2])
chr<-final2[,1]

pns<-clusterMaker(chr, position, order.it=TRUE, maxGap=300)

mod<-model.matrix(~as.factor(pheno[,8]) + (pheno[,9]) + (pheno[,10]) + pheno[,11] +
as.factor(Sample_Group), data=pheno)
mod0<-model.matrix(~as.factor(pheno[,8]) + (pheno[,9]) + (pheno[,10]) + pheno[,11],
data=pheno)

mval4<-data.matrix(mval3)
test<-dmrFind(logitp=mval4, mod=mod, mod0=mod0, coeff=6, pns=pns, chr=chr, pos=position)

## TYPE test$dmrs for dmrs

##WGCNA CODE IS FREELY AVAILABLE AT
http://labs.genetics.ucla.edu/horvath/CoexpressionNetwork/Rpackages/WGCNA/. Parameters
used in the application of this package are explained in detail in the Supplemental Materials.
```

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